AN INTRODUCTION TO

# CONGENITAL HEART DISEASE

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# FOREWORD

BARAGWANATH HOSPITAL was opened ten years ago. It now has 1,800 beds with the specialities of Medicine and Surgery well established. Cardiology is centred around the work of the Cardiac Clinic and its associated unit for catheterization. The turnover of more than a quarter of a million new patients a year provides a very fine experience of unselected medicine for the undergraduate and postgraduate student. The opportunities for research are outstanding and have stimulated some notable contributions to medical literature.

With the increase in knowledge and the progress in surgical treatment in recent years it has become important for the student and practitioner to recognize the commoner congenital heart lesions. The complexities and finer details found in larger publications tend to make the student feel that the understanding of congenital heart disease is beyond his ability. A concise manual on this subject has thus become a matter of increasing necessity and the authors of this book have succeeded admirably in providing a presentation readily understandable to the student. Although this book does not provide a complete and detailed knowledge of congenital heart disease, it teaches basic principles and crystallizes the important clinical syndromes with a clarity and directness that will simplify the difficulties of the student and practitioner.

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*August* 1959.

# PREFACE

THE notable advances that have been made in the field of congenital heart disease in recent years, particularly the advent of open heart surgery, have created a considerable upsurge of interest in this subject. What was previously a museum curiosity, or an obscure syndrome known only to the more erudite student, has now become a problem of great practical importance as accurate diagnosis and assessment may lead to surgical correction of congenital deformities hitherto believed irremediable. Congenital heart disease, which has always provided a diagnostic challenge to the physician, has now also come within the domain of the student and practitioner.

This book does not claim to be a complete or comprehensive treatise on the subject of congenital heart disease. It is written primarily as a guide for the student and practitioner, and the aim has been to present the subject matter in a clear and concise form. Thus, only the commoner congenital anomalies are stressed and technical procedures such as cardiac catheterization and angiocardiography are but briefly mentioned. No classification of con-

[illegible]

stone to the fuller and more detailed study of this most important branch of medical science.

We wish to express our sincere thanks to the Departments of Medicine, Paediatrics and Radiology, Baragwanath Hospital, for access to records; to Mr L Fatti for the X-ray plates of pulmonary stenosis and total anomalous pulmonary venous drainage (Figs. 61
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to Miss L. Hirsowitz for assistance with the proof reading, and to Dr Louis Hirsowitz for his helpful and constructive criticism.

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Johannesburg,
*August* 1959.

CHAPTER I

# THE APPROACH TO THE DIAGNOSIS OF CONGENITAL HEART DISEASE

It is the bizarre that often draws attention to the presence of congenital heart disease. The striking feature may be a pronounced thrill and murmur, the presence of severe cyanosis or associated congenital abnormalities elsewhere in the body. Gross signs are sometimes present with little disability.

## Aetiology of Congenital Heart Disease

The aetiology of congenital heart disease is usually obscure. Maternal rubella during the first 12 weeks of pregnancy has been shown to be a cause. Other viral infections such as measles, mumps, chickenpox, herpes zoster, infective hepatitis and poliomyelitis have likewise been incriminated. There is, however, no conclusive evidence to support this (Annot., *Brit. med. J.*, 1958).

## Associated Congenital Anomalies

Congenital heart disease may be associated with other visceral or skeletal malformations:

1. *Arachnodactyly* (spider fingers). In this condition the fingers are abnormally long and thin (Fig. 1).
2. *Marfan's Syndrome.* The combination of arachnodactyly with other skeletal deformities and abnormalities of connective tissue is known as Marfan's syndrome (Marfan, 1896). The features of this syndrome include long slender limbs (Fig. 1), poor muscle development, lack of subcutaneous tissue, laxity of ligaments, [illegible]

myopia, strabismus, nystagmus).

Atrial septal defect is the commonest congenital cardiac anomaly associated with Marfan's syndrome.

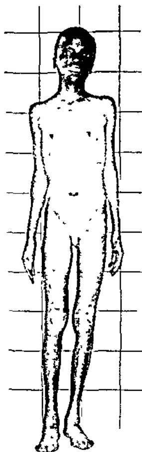


FIG 1 A case of Marfan's syndrome *Note* (*a*) long slender limbs, (*b*) arachnodactyly In addition there is associated hypertelorism—widely spaced eyes

3. *Hypertelorism.* This is a craniofacial deformity in which the eyes are widely spaced due to an increased breadth of the bridge of the nose. It may occur as an isolated congenital defect or with Marfan's syndrome (Fig. 1).
4. *Mongolism, chest deformities, mental deficiency* and *deafness:* These anomalies are occasionally found with congenital heart disease.

### Common Symptoms and Signs

*Breathlessness on exertion, fatigue* and *syncope* are particularly common in cyanotic congenital heart disease, e g. Fallot's tetralogy. Children often adopt a *squatting posture* to obtain relief from the dyspnoea.

The rate of growth and degree of physical development may reflect the functional severity of the anomaly. Thus, *stunted growth* and *failure to thrive* may occur with cyanotic congenital heart disease or large left to right shunts.

*Recurrent attacks of pulmonary infection* are common in malformations associated with plethoric or congested lungs.

*Cyanosis* is a characteristic feature of *certain forms of congenital heart disease,* e.g. Fallot's tetralogy, transposition of the great vessels and persistent truncus arteriosus. It may be central (tongue and extremities) or peripheral (extremities only) (see Chapter 3). In patent ductus arteriosus with reversal of blood flow, i.e. from pulmonary artery to aorta, cyanosis is more marked in the lower limbs.

*Clubbing of the fingers and toes* is a salient feature of all forms of cyanotic congenital heart disease. Cyanosis always precedes the development of clubbing.

The *character of the pulses* is often of great diagnostic value, e g. the pulses are weak with pulmonary stenosis and collapsing with patent ductus arteriosus Strong pulsations in the upper limbs with weak



FIG 2 Showing a high-arched palate (same case as Fig 1)

and delayed pulsations in the lower limbs are typical of coarctation of the aorta.

*Deformity* [illegible] of the right ventric[illegible] hyper-trophied rig[illegible]ic cage results in anterior bulging of the precordium. It may occur with congenital or acquired heart disease.

*Cardiac murmurs* associated with congenital heart disease are usually striking. The murmurs are frequently harsh and often accompanied by pronounced *thrills*. Murmurs may be heard in sites which are unusual for acquired heart disease. The *quality of the second heart sound* is of diagnostic import. A split second heart sound indicates closure of the pulmonary and aortic valves and is clear evidence of the existence of both great vessels. Thus, the second

sound is single and unsplit in atresia of either great vessel, severe forms of Fallot's tetralogy and persistent truncus arteriosus.

### Electrocardiography

The electrocardiogram is of value in the diagnosis of congenital heart disease. It may be decisive in determining which ventricle is dominant where clinical examination has failed. Congenital anomalies which shunt blood into the right side of the heart (e.g. atrial septal defect) or create a resistance to the outflow of the right ventricle (e.g. pulmonary stenosis) will be associated with right ventricular hypertrophy. Patent ductus arteriosus (in the absence of pulmonary hypertension), coarctation of the aorta and tricuspid atresia impose a strain on the left ventricle.

Enlargement of the atria may be demonstrated. Abnormally high and peaked P waves *(best seen in standard lead 2) occur with*

[illegible] interval

[illegible] Partial

or complete right bundle branch block is common with atrial septal defect.

Complete heart block may occur as an isolated congenital anomaly. It differs from acquired heart block in that the rate is often faster (40 to 80 beats per minute) and is increased by exercise and atropine, the QRS complexes are *always* normal and syncopal attacks are rare.

The electrocardiogram is characteristic in dextrocardia. The leads reveal a 'mirror-image' of the normal pattern (see Fig. 81, page 109)

The electrocardiogram is of particular value in the diagnosis of anomalous origin of the left coronary artery from the pulmonary artery. The characteristic patterns of myocardial infarction may be found, viz. deep wide Q waves, raised S-T segments and inverted T waves in leads facing the injured myocardium

### Radiology

Radiological examination is a valuable aid to diagnosis and may reveal the following information.

1. *The size of the heart and its chambers.*
2. *Unusual contours* e g , the 'coeur en sabot' or the 'upturned boot' of Fallot's tetralogy, the 'figure-of-eight' or 'cottage loaf' of total

anomalous pulmonary venous drainage, the narrow vascular pedicle of transposition of the great vessels and the wide vascular pedicle of persistent truncus arteriosus.

3. *The degree of cardiac pulsation*, e.g. the hyperactive heart of aortic incompetence associated with bicuspid aortic valves and the 'quiet' heart of Ebstein's anomaly.
4. *The character of the pulmonary arteries and lung fields.* If blood is

pulmonary arteries results in increased vascularity of the lung fields. If this volume of blood is greatly increased the pulmonary arteries become large and pulsatile (hilar dance).

The decreased flow of blood to the lungs in pulmonary stenosis and atresia is revealed by *diminished vascularity of the lung fields.*

5. *Unusual signs*, e.g. notching of the ribs in coarctation of the aorta.

## CARDIAC CATHETERIZATION

Cardiac catheterization is a complicated procedure It requires a team of at least four experienced persons and entails the use of intricate apparatus. It may be briefly described as follows·

Under local anaesthesia a catheter is inserted through an antecubital vein and passed into the right atrium, right ventricle and pulmonary arteries. Pressures are recorded in each of the above sites and blood samples are withdrawn for oxygen studies Blood oxygen studies, to be valid, demand a restful and sedated patient. Small children may require a general anaesthetic

The following observations and studies may be made during cardiac catheterization.

1. *Visualization of the catheter.* The catheter may be seen in an abnormal site due to its passage through a septal defect, patent ductus arteriosus or anomalous pulmonary vein.
2. *Oxygen studies.* These may reveal the presence of a shunt, e.g. the oxygen content of blood from the pulmonary arteries is increased in patent ductus arteriosus.
3. *Pressure studies.* The pressure in a chamber or vessel may be increased. An augmented pressure gradient between two chambers occurs when the communicating aperture is stenosed, e.g. in

pulmonary stenosis a high pressure is recorded in the right ventricle and a low pressure in the pulmonary artery.

4. *Angiocardiography.* This procedure is sometimes performed during cardiac catherization. It may elicit more precise information about the anatomical defect, e.g. the nature and site of the narrowing in pulmonary stenosis.

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Annotation (1958). [illegible]
Marfan, A. B. (1896) [illegible]
plus prononcée aux [illegible]
*certain degré d'amu* [illegible]

CHAPTER 2

# DEVELOPMENT OF THE HEART*

THE heart develops from a pair of symmetrical vessels called the *primitive heart tubes.* These vessels are the caudal continuation of the paired *ventral aortae* which are connected with the *dorsal aortae* through the *first aortic arches* (Fig. 3).

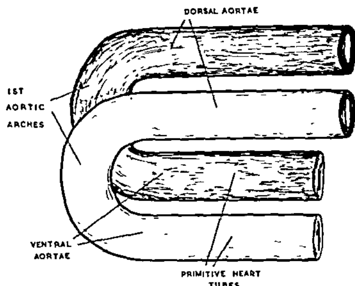



FIG 3 Lateral view of the primitive heart tubes and aortae

The primitive heart tubes fuse to form a single *primitive* tubular heart (Fig. 4).

* For convenience and clarity the embryology of the heart is related chronologically It must, however, be emphasized that many changes take place simultaneously and that for exact temporal relationships the reader is referred to more comprehensive texts on the subject

Two constrictions develop in this primitive tubular heart dividing it into three sacculations—the *bulbus cordis, ventricle* and *atrium* (Fig 5).

At the caudal end of the atrium a further sacculation develops—*the sinus venosus*. Similarly, the cephalic end of the bulbus cordis is constricted to form the *truncus arteriosus* (Fig. 6). The sinus venosus receives the venous return to the primitive heart and the truncus arteriosus forms the arterial outlet.

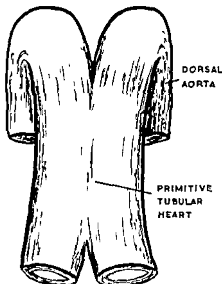
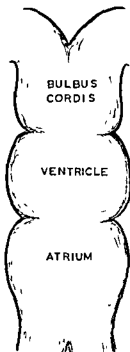



Fig 4 Ventral view showing fusion of the heart tubes to form a single primitive tubular heart

With rapid and unequal growth the heart is moulded into a spiral S-shaped tube (Figs. 7 and 8). The atrium and later the sinus venosus come to lie posterior to the bulbus cordis and ventricle thus forming an S-shaped tube in the sagittal plane (Figs. 7 *a* and 8). The main flexure of this S-shaped tube is also moulded to form a U-shaped tube in the coronal plane (Fig. 7 *b*). The combination of the S- and U-shaped tubes is thus in effect a spiral. The U-shaped tube, comprising the bulbus cordis and ventricle, is known as the *bulbo-ventricular loop*.

The atrium is prevented from expanding dorsally by the gut and



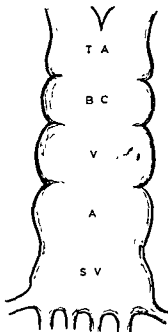
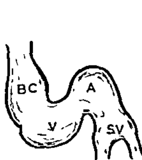




FIG 5 FIG 6

FIG 5 Ventral view of the primitive heart tube showing its division into bulbus cordis, ventricle and atrium

FIG. 6 Ventral view showing the five primary sacculations of the primitive heart tube T A, truncus arteriosus, B C, bulbus cordis, V, ventricle, A, atrium, S V, sinus venosus



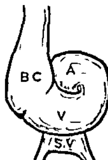




FIG 7 Showing formation of the spiral S-shaped heart tube (a—sagittal view, b—ventral view) B C, bulbus cordis; V., ventricle, A, atrium, S V., sinus venosus

ventrally by the bulbus cordis. Growth therefore occurs laterally, the atrium bulging on either side of the bulbus cordis (Fig. 9). These lateral expansions are the future right and left atria.

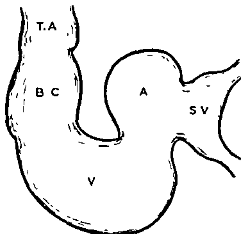



FIG. 8. Sagittal view of the S-shaped heart tube showing the relationship of its chambers T A, truncus arteriosus, B C, bulbus cordis, V, ventricle, A, atrium, S V, sinus venosus

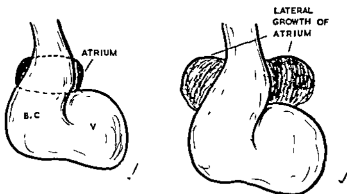



FIG. 9 Ventral view of the heart showing lateral growth of the atrium B C, bulbus cordis, V, ventricle.

As the bulbo-ventricular loop grows the septum or bulbo-ventricular fold, which demarcates the bulbus cordis and ventricle,

disappears. These two compartments merge into a single chamber which is the precursor of the right and left ventricles (Fig. 10).

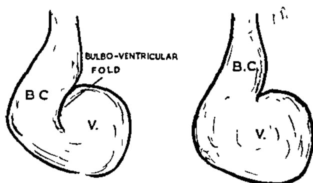



FIG. 10 Showing disappearance of the bulbo-ventricular fold and the formation of a single ventricular chamber B C, bulbus cordis, V, ventricle

## DEVELOPMENT OF THE FOUR-CHAMBERED HEART

The atrium and ventricle communicate through the *atrio-ventricular canal.* Partition of this canal is effected by dorsal and ventral

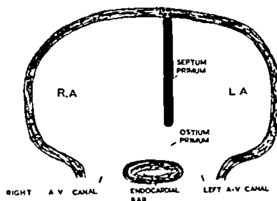



FIG 11 Transverse section of the primitive atrium showing its division into right and left atria by the septum primum A-V., atrio-ventricular; R.A, right atrium, L.A., left atrium.

### DEVELOPMENT OF THE ATRIA

The single primitive atrium is divided into left and right atria by the downward growth from its dorsal wall of a sickle-shaped membrane—the *septum primum* (Fig. 11). The free edge of this septum fuses with the endocardial bar (Fig. 12). A localized area of the septum primum becomes thinned and perforate forming a communication between the right and left atria—the *foramen ovale I* (Fig. 12).

A second membrane—the *septum secundum*—now develops from the dorsal wall of the atrium on the right side of the septum primum.

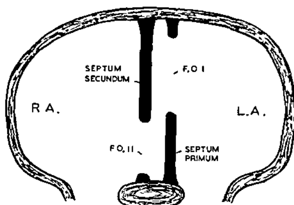



FIG 12 Transverse section of the primitive atrium showing the formation of the septum secundum and foramina ovale R A, right atrium, L A, left atrium, F O. I, foramen ovale I, F O II, foramen ovale II

This membrane is incomplete and overlaps the foramen ovale I The defect in the septum secundum is termed the foramen ovale II (Fig. 12) The foramina ovale I and II are sometimes collectively referred to as the *foramen ovale.* The septum primum and septum secundum fuse at birth to form the interatrial septum.

### DEVELOPMENT OF THE VENTRICLES

Reference has already been made to the incorporation of part of the bulbus cordis into the ventricle to form a single chamber (Fig. 10). This single ventricle is then divided into two compartments by the *interventricular septum.* This septum consists of two

parts—the *muscular* and the *membranous* interventricular septa. The muscular interventricular septum develops as an inward projection from the floor of the primitive ventricle and grows towards the endocardial bar (Fig. 13). Simultaneously, the spiral septum, which separates the primitive aorta and pulmonary artery (see page 14), develops from endocardial ridges in the truncus arteriosus. The muscular interventricular septum, spiral septum and endocardial

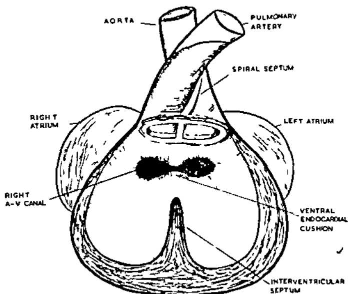



FIG 13 Semidiagrammatic representation showing development of the ventricles, pulmonary artery and aorta

bar do not fuse. This results in a small opening—the *interventricular foramen*—which is subsequently closed by the growth of a membranous septum. This septum, which forms the membranous part of the interventricular septum, grows downwards from the endocardial ridges of the truncus arteriosus and upwards from the endocardial bar.

## Development of the Sinus Venosus and Great Veins

This is discussed in Chapter 14, page 101.

## Development of the Aorta and Pulmonary Artery

The tubular bulbus cordis and truncus arteriosus are divided by a septum—the *bulbar or spiral septum*—into the roots of the aorta and pulmonary artery. This septum develops from endocardial ridges which grow from the wall of the truncus arteriosus and bulbus cordis and fuse in the midline. It spirals caudally so that the pul-

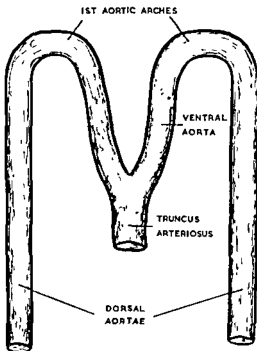



Fig 14 Showing paired dorsal and ventral aortae connected by the first aortic arches

monary artery lies ventral to the aorta and opens into the right ventricle, while the aorta opens into the left ventricle (Fig 13).

The aortic arch and main branches of the aorta develop from the primitive paired ventral and dorsal aortae. These vessels are connected at their cephalic ends by the first aortic arches (Figs. 3, 4, 14).

Further vessels develop between the dorsal and ventral aortae to

form a total of six paired aortic arches The fifth aortic arch is rudimentary and its existence has been disputed. The dorsal aortae fuse caudally to form the descending aorta (Fig 15).

The first and second arches disappear. The dorsal aortae at the level of the first three arches together with the third aortic arches

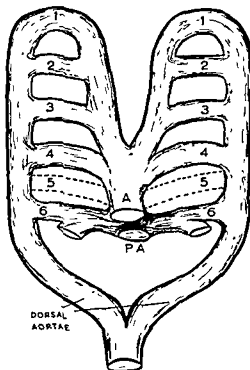



Fig 15 Dorsal view of the six primitive paired aortic arches. P A, pulmonary artery, A, aorta

form the internal carotid arteries. The ventral aortae persist as the external carotid arteries (Fig. 16) The dorsal aortae between the third and fourth arches are obliterated The remainder of the right dorsal aorta also disappears The right fourth aortic arch becomes the right subclavian artery. The left fourth aortic arch and the remaining part of the left dorsal aorta form the aortic arch proper. An outgrowth from this arch forms the left subclavian artery. The

right sixth arch disappears. The left sixth arch persists as the ductus arteriosus (Fig. 16. See also section on embryology in Chapter 4).

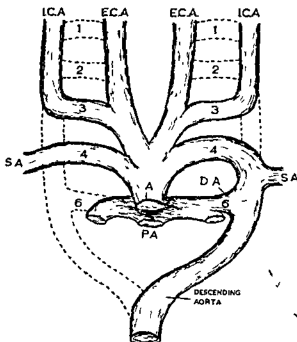



FIG 16 Dorsal view of the aortic arches showing formation of the principal vessels A, aorta, P A, pulmonary artery, D A, ductus arteriosus, S A, subclavian artery, E C A, external carotid artery, I C A, internal carotid artery

## THE FOETAL CIRCULATION

*venosus* (a large vessel which lies on the posterior surface of the liver) into the *inferior vena cava* A small amount of blood is shunted through the liver itself The inferior vena cava also receives un-

limbs. Both vena cavae enter the right atrium.

Blood from the right atrium flows through two channels, viz. (*a*) through the *foramen ovale* to the left atrium; (*b*) through the tricuspid valve to the right ventricle. From the right ventricle blood is directed into the *pulmonary artery*. The greater part of this

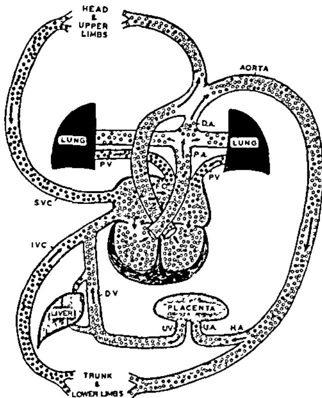



FIG 17 The foetal circulation. D A, ductus arteriosus, P A, pulmonary artery, P V, pulmonary vein, F O, foramen ovale, S V C, superior vena cava, I V C, inferior vena cava, D V, ductus venosus, H A, hypogastric artery, U A, umbilical artery, U V, umbilical vein. Black circles denote unsaturated blood, white circles denote saturated blood

flow passes through the *ductus arteriosus* into the aorta and thence to the trunk and lower limbs. A small volume of the pulmonary artery flow goes to the functionless lungs via the right and left pulmonary arteries and returns to the left atrium through the

pulmonary veins. The left atrium receives a small amount of [illegible]

ventricle and *aorta* and is directed to the head and upper limbs. [illegible]

the descending aorta passes to the placenta by way of the *hypogastric and umbilical arteries.*

## CHANGES IN THE CIRCULATION AT BIRTH

During foetal life the right ventricle has to pump against the high resistance of the collapsed and functionless lungs. It also has to pump blood through the ductus arteriosus to the lower limbs There is thus a high pressure in the right ventricle and pulmonary artery.

At birth, with aeration of the lungs, a fall in pressure occurs in the pulmonary circulation (see also page 41, Figs. 37, 38). The pressure in the pulmonary artery now approximates that in the aorta With equalization of pressures in the pulmonary artery and aorta the flow through the ductus arteriosus ceases and all the blood in the pulmonary artery is directed to the lungs. The increased pulmonary circulation results in a greater return of blood to the left atrium. The pressure in the left atrium rises and leads to closure of the foramen ovale.

## CHAPTER 3

# CYANOSIS

CYANOSIS is the blue or violet colour of the skin and mucous membranes which occurs when the surface capillaries contain an excess amount of reduced haemoglobin. Rarely, it is due to abnormal haemoglobin derivatives such as sulphaemoglobin and methaemoglobin.

At least 5 grams reduced haemoglobin per 100 c c. of blood must be present in the *capillary circulation* for the development of cyanosis (Lundsgaard, 1919) (Figs. 18 and 19).

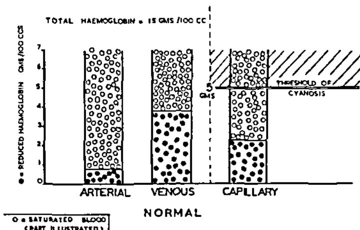



FIG 18A Showing the relative amounts of reduced haemoglobin in arterial, venous and capillary blood under normal conditions. *Note* The amount of reduced haemoglobin in capillary blood is the mean of the arterial and venous levels

With normal conditions and a haemoglobin of 15 grams per 100 c.c. *arterial blood* contains approximately 0 75 gram reduced haemoglobin per 100 c.c (Fig. 18A) Under similar conditions,

*venous blood* contains approximately 3·75 grams reduced haemoglobin per 100 c.c. (Fig. 18A).

The amount of reduced haemoglobin in the *capillary blood* is the mean of the arterial and venous values:

GRAMS REDUCED HAEMOGLOBIN PER 100 C.C. OF BLOOD

Example (Fig. 18A)

$$\frac{\textit{Arterial}\ 0{\cdot}75 + \textit{Venous}\ 3{\cdot}75}{2} = \textit{Capillary}\ 2{\cdot}25$$

Thus, under normal conditions the capillaries contain approximately 2·25 grams reduced haemoglobin per 100 c.c. This figure

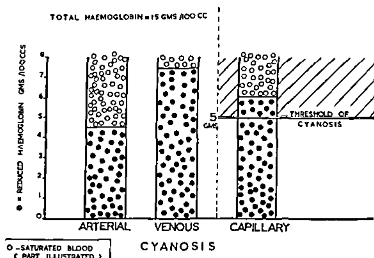



Fig. 18B Showing an example of the relative amounts of reduced haemoglobin in arterial, venous and capillary blood in cyanosis. The cause of the cyanosis in this instance is decreased arterial oxygenation

is well below the minimum threshold for the development of cyanosis.

Cyanosis is due to an increased amount of reduced haemoglobin in the capillaries and is dependent on the following factors:

1.

2.

3.

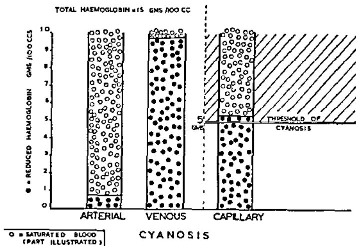



FIG 18C Showing an example of the relative amounts of reduced haemoglobin in arterial, venous and capillary blood in cyanosis The cause of the cyanosis in this instance is an increased arterio-venous oxygen difference

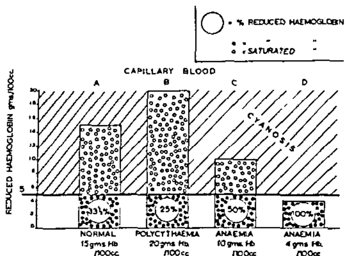



FIG 19 Showing the percentage reduced haemoglobin required to attain the *minimum* threshold level for cyanosis with different total haemoglobin concentrations.

## 1. THE AMOUNT OF REDUCED HAEMOGLOBIN IN THE ARTERIAL BLOOD

An increased amount of reduced haemoglobin in the arterial blood must inevitably result in a proportionate increase of reduced haemoglobin in the capillary and venous blood:

GRAMS REDUCED HAEMOGLOBIN PER 100 C.C. OF BLOOD

| Example (FIG. 18B) | *Arterial* | | *Venous* | | *Capillary* |
|---|---|---|---|---|---|
| | 4·5 | + | 7·5 | | |

$$\frac{4{\cdot}5 + 7{\cdot}5}{2} = 6$$

This amount of reduced haemoglobin in the capillary blood—6 grams per 100 c.c.—is above the minimum threshold for cyanosis and cyanosis thus appears.

An increased amount of reduced haemoglobin in the arterial blood may result from:

(*a*) Veno-arterial shunts.
(*b*) Impaired arterial oxygenation.

### (*a*) *Veno-arterial shunts*

In right to left shunts (e.g. Fallot's tetralogy, Eisenmenger's syndrome) venous blood is shunted from the right side of the heart and mixes with arterial blood in the left side of the heart and systemic circulation. As a result, part of the systemic venous return to the heart by-passes the lungs and is not available for oxygenation (Fig. 20). In addition, the admixture of venous and arterial blood causes an increased amount of reduced haemoglobin in the arterial circulation (Fig. 20).

### (*b*) *Impaired arterial oxygenation*

This may result from:

(i) Impaired diffusion of oxygen due to pulmonary disease, e.g. consolidation, atelectasis, emphysema, fibrosis.
(ii) A low partial pressure of the alveolar oxygen as occurs at high altitudes.

When cyanosis is due to impaired arterial oxygenation, the inhalation of oxygen will improve the arterial oxygen saturation and

alleviate the cyanosis. Oxygen inhalation, however, has *no* effect on the cyanosis due only to a right to left shunt.

## 2. THE DEGREE OF UTILIZATION OF CAPILLARY OXYGEN BY THE TISSUES

Gaseous exchange occurs in the capillaries. Increased utilization of oxygen by the tissues results in an increased amount of reduced

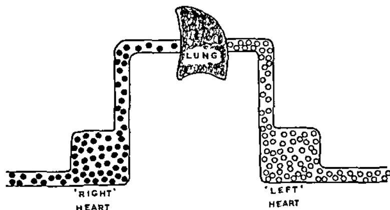
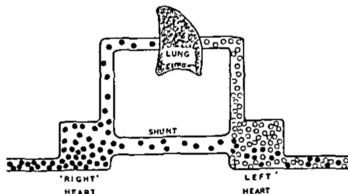



FIG 20A. Diagrammatic representation of the normal pulmonary circulation Black circles denote unsaturated blood, white circles denote saturated blood



haemoglobin in the venous blood. Thus, the amount of haemoglobin converted to reduced haemoglobin during passage through the capillaries is greater than normal, i.e. there is an *increased arterio-venous oxygen difference*

GRAMS REDUCED HAEMOGLOBIN PER 100 C.C OF BLOOD

| Example (FIG. 18C) | *Arterial* | | *Venous* | | *Capillary* |
|---|---|---|---|---|---|
| | 0·75 | + | 9·75 | | |
| | | 2 | | = | 5·25 |

This amount of reduced haemoglobin in the capillary blood—5·25 grams per 100 c.c.—is slightly above the minimum threshold for cyanosis.

Any condition which results in a sluggish peripheral circulation (e.g. congestive cardiac failure, shock, exposure to cold) may cause cyanosis due to this mechanism The degree of cyanosis is usually mild. It can be appreciated, however, that when the amount of reduced haemoglobin in the arterial blood is already elevated from other causes, any increase in arterio-venous oxygen difference will aggravate or precipitate cyanosis.

### 3. THE TOTAL HAEMOGLOBIN CONCENTRATION

The percentage reduced haemoglobin required to attain the minimum threshold for cyanosis (5 grams per 100 c.c. of blood) depends on the total haemoglobin concentration. Thus

1. With a normal haemoglobin of 15 grams per 100 c.c. of blood, at least 33⅓ per cent of the haemoglobin must be in reduced form for the development of cyanosis (Fig. 19, column A).
2. With polycythaemia (e.g. 20 grams haemoglobin per 100 c.c. of blood), only 25 per cent of the haemoglobin need be in reduced form for the development of cyanosis (Fig. 19, column B)
3.

of *all* the circulating haemoglobin being in reduced form (Fig. 19, column D).

## Distribution of Cyanosis

Cyanosis may be *central* or *peripheral*.

*Central cyanosis* is due to a central mechanism, i.e. diminished arterial oxygen saturation It is visible in the skin (nose, cheeks, fingers) and in warm areas, viz. those richly supplied with blood vessels (tongue, lips, conjunctivae). Central cyanosis, if severe and long lasting, is associated with clubbing of the fingers and polycythaemia.

*Peripheral cyanosis* is due to a peripheral mechanism, i e an
It is visible only in the
rm areas (tongue, lips,
polycythaemia do not
occur.

REFERENCE

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## CHAPTER 4

# COARCTATION OF THE AORTA

### Anatomy

Coarctation (Latin *cum*—together, and *arcta're*—to make tight) of the aorta is a congenital anomaly in which there is a stricture or narrowing of the aorta. It may occur at any site along the course of the aorta but is most commonly found just below, or in the

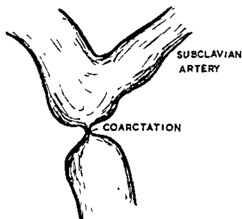



Fig 21 Showing the localized and abrupt form of coarctation of the aorta

site of the coarctation.

### Associated Anomalies

The congenital malformations most commonly associated with

coarctation of the aorta are *patent ductus arteriosus* and *bicuspid aortic valve*. Less frequent concomitant anomalies are aneurysms of the circle of Willis and subaortic stenosis. Rarely, extravascular malformations such as double vagina, undescended testis and ectopic and polycystic kidneys may be found.

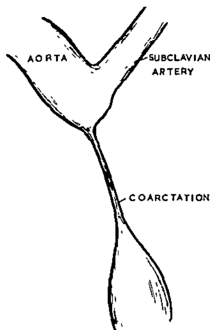



FIG. 22 Showing the elongated form of coarctation of the aorta

## EMBRYOLOGY

The aorta develops from the primitive aortae and the fourth aortic arch on the left side (see also Chapter 2 and Figs. 14, 15, 16, 23) The ductus arteriosus is formed from the left sixth aortic arch and joins the left dorsal aorta between the fourth aortic arch and the point of fusion of both dorsal aortae. This region of the aorta, i.e just above and below the ductus arteriosus, is the commonest site for coarctation of the aorta (Fig. 23).

## Dynamics of the Circulation

Cases of coarctation of the aorta may be classified into two types* (Kjellberg *et al.*, 1955):

1. Coarctation of the aorta *not* associated with a functionally significant ductus arteriosus. The ductus arteriosus is usually completely obliterated If the ductus arteriosus remains patent,

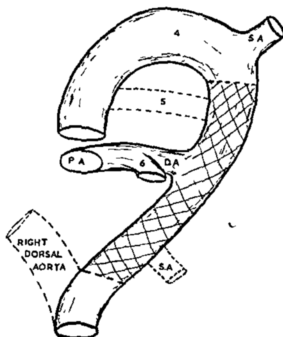



Fig 23 Diagrammatic representation showing development of the aorta The shaded area denotes the commonest site for coarctation of the aorta P A, pulmonary artery, D A, ductus arteriosus, S A, subclavian artery Note The left subclavian artery arises from the left dorsal aorta distal to the entrance of the ductus arteriosus and due to unequal growth of the aorta migrates in a cephalic direction

the flow is minimal and the direction of the shunt is always from *aorta to pulmonary artery* irrespective of whether the ductus opens above or below the constriction. This is the common type of coarctation.

* The terms 'adult' coarctation (a constriction below the ductus arteriosus) and 'infantile' coarctation (a constriction above the ductus arteriosus) are misleading and should be abandoned

2. Coarctation of the aorta associated with a functionally significant ductus arteriosus. The flow through the ductus arteriosus is from pulmonary artery to aorta. The direction of flow is thus similar to that found in the foetal circulation and is the reverse of that occurring in an uncomplicated patent ductus arteriosus. Consequently, the lower half of the body receives venous blood

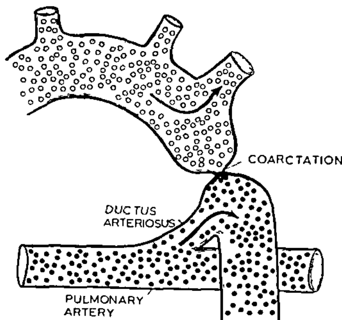



FIG 24 Coarctation of the aorta with a functionally significant ductus arteriosus
Note The shunt through the ductus arteriosus is from pulmonary artery to aorta
Black circles denote unsaturated blood; white circles denote saturated blood

from the pulmonary artery *via* the ductus arteriosus (Fig 24). As a result, the right ventricle has to pump against the high resistance of the systemic circulation. This places a strain on the right ventricle and death usually occurs in early infancy. The clinical features are those of right ventricular failure and cyanosis in the lower extremities.

The remainder of this chapter is concerned only with the common type of coarctation of the aorta, viz. *coarctation of the aorta without a functionally significant ductus arteriosus.* In this form the circulation

to the lower half of the body occurs via collateral vessels. These channels arise from the subclavian artery or its branches and anastomose with the aorta or its branches distal to the constriction. There are three systems of collateral vessels:

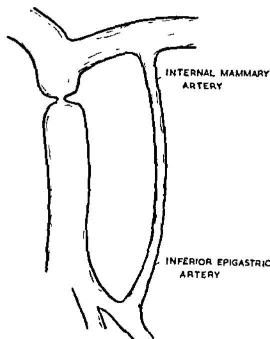



FIG 25 Semidiagrammatic representation of the collateral channel formed by the internal mammary and inferior epigastric arteries

1. The internal mammary artery (branch of the subclavian artery) anastomoses with the inferior epigastric artery (branch of the external iliac artery) (Fig. 25).
2. The anastomosis around the shoulder girdle. This is effected by the transverse cervical and transverse scapular arteries (branches of the thyrocervical trunk) and the subscapular artery (branch of the axillary artery). These vessels form a circumscapular anastomosis. Vessels from this network pierce the posterior intercostal spaces and anastomose with the second, third and fourth intercostal arteries (branches of the aorta) (Fig. 26).

3. The superior intercostal artery (branch of the costocervical trunk) anastomoses with the first intercostal artery (branch of the aorta) (Fig. 27).

## Age Incidence

Most patients do not live beyond the age of 50 years Survival up to the age of 92 years has, however, been reported (Abbott, 1928).

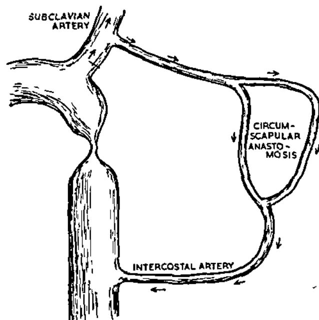



*Fig 26 Semidiagrammatic representation of the collateral channel* formed by the circumscapular anastomosis

## Sex Incidence

Coarctation of the aorta is more common in males.

## Clinical Features

Patients with coarctation of the aorta are well developed physically. The lower limbs may be small in proportion to the rest of the body (Fig. 28).

Prominent pulsation of the carotid arteries may be evident. This is especially marked when there is associated aortic incompetence due to a bicuspid aortic valve.

Pulsations in the femoral arteries are weak and sometimes impalpable. When palpable, the femoral pulsations are *delayed* as compared with the radial pulses. Visible or palpable collateral vessels may be detected in the interscapular region. These are best

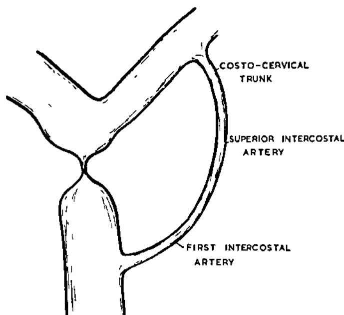



FIG 27. Semidiagrammatic representation of the collateral channel formed by the superior intercostal and first intercostal arteries

observed when the patient bends forward with the arms dependent (Campbell and Suzman, 1947)

A most important sign is the presence of hypertension in the upper limbs. The blood pressure in the legs is lower than in the arms. This does not necessarily imply a normal or hypotensive level in the legs. Despite the difference in blood pressure between the arms and legs, the blood pressure in the legs may still reach hypertensive levels. If the coarctation involves the left subclavian artery, the blood pressure in the left arm will be lower than that in the right arm.

There is mild or moderate cardiac enlargement. Gross cardiomegaly usually indicates a complicating factor, most commonly aortic incompetence due to a bicuspid aortic valve. The apical cardiac impulse is characteristic of left ventricular hypertrophy, viz. localized and forceful.

On auscultation there is a loud early systolic murmur best heard in the aortic area and radiating into the neck and mitral regions. This murmur is often initiated by a sharp early systolic sound—an ejection click (Fig. 29). In addition, a late systolic murmur may be present over the site of the constriction This murmur continues up to the second heart sound and may spill over into diastole. It is loudest posteriorly between the scapulae but may also be heard anteriorly over the second and third intercostal spaces to the left of the sternum (Fig. 30) An apical mid-diastolic murmur is heard intermittently in a third of cases (Fig. 29). Its cause is still speculative The presence of an early diastolic murmur indicates aortic incompetence which is most often due to an associated bicuspid aortic valve (Fig. 31).

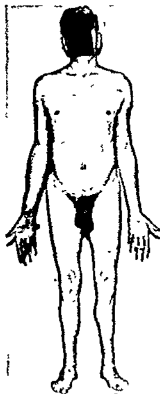


FIG 28 Patient with coarctation of the aorta Note the well developed build and the disproportionately small lower limbs

## ELECTROCARDIOGRAPHY

The electrocardiogram often shows left ventricular hypertrophy, viz. deep S waves in right ventricular leads (usually V1 and V2) and tall R waves in left ventricular leads (usually V5 and V6). In addition, left ventricular strain may be present as revealed by inverted T waves and depressed S-T segments in left ventricular leads (Fig. 32). Marked left ventricular hypertrophy and strain is most common in cases complicated by aortic valve disease.

NORMAL

IST HS 2ND HS

A P

1 EC SM 2 A P

1 SM 2 A P MDM. 30 % OF CASES

Fig 29 Auscultatory findings over the anterior chest wall in coarctation of the aorta H S, heart sound, A, aortic element of second heart sound, P, pulmonary element of second heart sound, E C, ejection click, S M, systolic murmur, M D M, mid-diastolic murmur, 1, first heart sound, 2, second heart sound

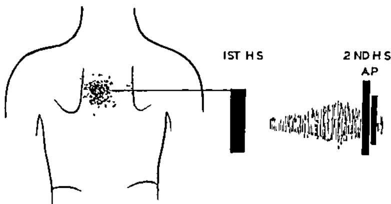



Fig 30 Auscultatory findings over the interscapular region in coarctation of the aorta H S, heart sound, A, aortic element of second heart sound, P, pulmonary element of second heart sound.

NORMAL
1ST H S
2ND H S
A P
1 E C
S M
2
A P
E D M
1
S M
2
A P
M D M 30 %
OF CASES

Fig 31 Auscultatory findings in coarctation of the aorta associated with bicuspid aortic valve H S, heart sound, A, aortic element of second heart sound, P, pulmonary element of second heart sound, E C, ejection click; S M, systolic murmur, E D M, early diastolic murmur, M D M, mid-diastolic murmur, 1, first heart sound, 2, second heart sound

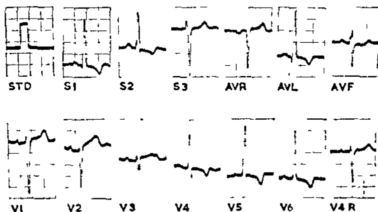



Fig 32 Electrocardiogram showing left ventricular hypertrophy and strain (deep S waves in leads V1 and V2, tall R waves in leads V5 and V6, inverted T waves and depressed S–T segments in leads V4, V5 and V6)

## Radiology

The heart may be normal in size or show evidence of left ventricular hypertrophy. Gross left ventricular enlargement is usually associated with aortic incompetence (Figs 33, 34). *The postero-anterior view of the chest often shows a dilated left subclavian artery* (A in Fig 33) which may be mistaken for the aortic knuckle. A more penetrated film, however, will differentiate the aortic knuckle

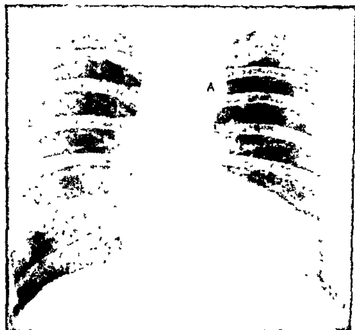



Fig 33 X-ray of chest in coarctation of the aorta showing (1) gross left ventricular enlargement due, in this patient, to an associated bicuspid aortic valve, (2) dilatation *of the left subclavian artery giving the appearance of a false aortic knuckle*— (A), (3) notching of the inferior margins of the ribs posteriorly

from the subclavian artery. The true aortic knuckle (B in Fig. 34) is seen as a double density below the subclavian artery (A in Fig. 34)

A striking feature is the presence of *notching of the ribs*. It involves the inferior margins of the ribs posteriorly and is due to pressure from the dilated intercostal arteries (Figs. 33, 34, 35) It is rarely observed in young children.

The aorta immediately distal to the stricture may be dilated—post-stenotic dilatation. The coarctation itself may sometimes be visualized in the oblique and postero-anterior views and may also be demonstrated by retrograde aortography.

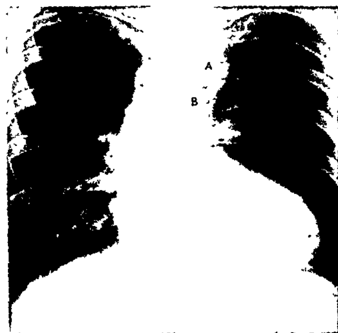



Fig 34 Penetrated view of the chest (same patient as in Fig 33) Note the dilated left subclavian artery (A) and the aortic knuckle (B)

## Clinical Presentation

The patient may present in the following ways.

1. There are no symptoms and the condition is detected on routine examination, e.g life insurance examination.
2. The patient complains of *headache*, *dizziness*, *tinnitus*, *epistaxis*, *palpitations* and *insomnia* due to the hypertension and the increased circulation to the head and upper limbs.
3. The patient has symptoms due to the diminished circulation in the lower limbs, e.g *cold feet*, *intermittent claudication* and *delayed healing of wounds*

4. The patient may have symptoms caused by COMPLICATIONS.
   (a) *Cardiac failure* may occur but is uncommon in children and young adults unless there is a complicating factor such as aortic incompetence or subacute bacterial endocarditis.
   (b) *Rupture of the aorta* may result in sudden death. It is particularly prone to occur during parturition.



FIG 35 Showing notching of the inferior margins of the ribs posteriorly (enlarged section of Fig 33)

   (c) *Subacute bacterial endocarditis* may affect a bicuspid aortic valve or the aorta at the site of the coarctation. Involvement of a bicuspid aortic valve is more common.
   (d) *Cerebral haemorrhage* may result from hypertension or rupture of an associated cerebral aneurysm.

## TREATMENT

Conservative therapy includes the prophylaxis of subacute bacterial endocarditis (antibiotic cover during infections, puerperium and surgical procedures, especially dental surgery) and treatment of

the hypertension with hypotensive drugs Patients should be advised to avoid undue strain.

Owing to the risk of aortic rupture during parturition, delivery should be performed by caesarean section.

Since the advent of successful surgery the prognosis has improved considerably Surgical correction is recommended in all cases except the very mild. It is best performed between the ages of 10 and 20 years (Gross, 1953). After the age of 40 years surgery is hazardous and usually contraindicated.

## General Comment

The necessity for routine examination of the femoral arteries must be stressed. It is emphasized that a *delay* in femoral arterial pulsation, compared with the radial arterial pulsation, is as important as diminished femoral pulsation.

The blood pressure in the lower limbs should be recorded in every patient with hypertension and in all pregnant women at the first ante-natal examination.

The diagnosis of bicuspid aortic valve cannot be made with certainty on clinical examination. Its presence, when associated with coarctation of the aorta, is suggested by the finding of gross cardiomegaly and aortic incompetence.

The combination of hypertension and aortic incompetence should arouse suspicion of the possible presence of coarctation of the aorta

Aortic incompetence associated with coarctation of the aorta may be due to a bicuspid aortic valve or dilatation of the ascending aorta which results from the increased pressure and blood flow. Infection of a bicuspid aortic valve by bacterial endocarditis leads to distortion or rupture of the cusps and may thus aggravate or precipitate aortic incompetence.

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## CHAPTER 5

# PATENT DUCTUS ARTERIOSUS

### ANATOMY

THE ductus arteriosus is a short vessel which joins the root of the left pulmonary artery to the aorta immediately distal to the left subclavian artery (Fig 36) It forms an important circulatory channel in intrauterine life (*vide infra*). This vessel normally becomes functionally insignificant at birth and undergoes obliteration soon after. Its persistence as a patent vessel constitutes the congenital malformation of patent ductus arteriosus.

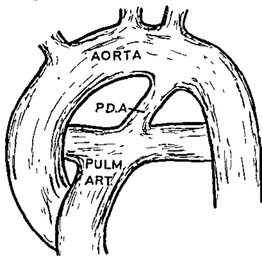



FIG 36 Showing anatomy of the ductus arteriosus P D A , patent ductus arteriosus, PULM ART , pulmonary artery.

### ASSOCIATED ANOMALIES

Patent ductus arteriosus usually occurs as an isolated abnormality. It may, however, be associated with other cardiovascular anomalies,

e g. coarctation of the aorta. It sometimes serves as an important compensatory channel, e g. when associated with Fallot's tetralogy, tricuspid atresia or transposition of the great vessels

## Embryology

The ductus arteriosus develops from the left sixth aortic arch and joins the primitive left dorsal aorta below the fourth aortic arch (refer Chapter 2 and Figs 16 and 23).

## Dynamics of the Circulation

During foetal life the lungs are unaerated and functionless Consequently, the pulmonary vascular resistance is high and the blood flow through the lungs is impeded The pressure in the

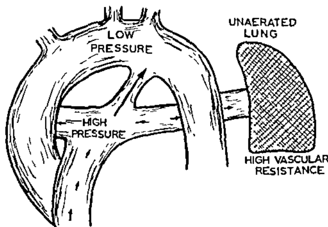



Fig. 37 Showing direction of blood flow through the ductus arteriosus *during foetal life*

pulmonary artery is higher than that in the aorta. Thus, the greater proportion of blood which is pumped into the pulmonary artery passes through the ductus arteriosus into the aorta (Fig. 37)

At birth, aeration of the lungs is followed by a rapid fall in pulmonary vascular resistance and the pressure in the pulmonary artery becomes less than that in the aorta. Consequently, if the ductus arteriosus remains patent, the flow through the ductus will be from aorta to pulmonary artery (Fig. 38) thereby increasing the

total pulmonary blood flow. This results in dilatation of the pulmonary artery and its branches and increases the load on the left atrium and left ventricle.

In a small percentage of cases the pulmonary vascular resistance remains high after birth. The pulmonary arterial pressure may then

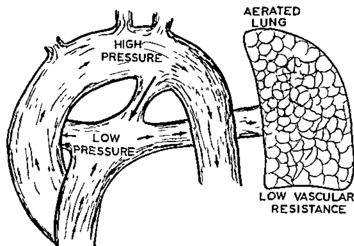



FIG 38 Showing direction of blood flow through the ductus arteriosus after birth

equal or exceed that in the aorta (see Chapter 9). The circulatory dynamics resulting from this state are discussed in this chapter under the heading—*Patent ductus arteriosus with pulmonary hypertension.*

### Age Incidence

Patent ductus arteriosus is usually diagnosed during the first and second decades It is rarely seen after the age of 60 years.

### Sex Incidence

Patent ductus arteriosus is more common in females

### Clinical Features

The peripheral pulses are collapsing or 'water-hammer' in quality. The pulse pressure is increased and the greater the shunt, the lower the diastolic blood pressure. A fall in diastolic blood pressure occurs with exercise—Bohn's sign (Bohn, 1938).

The heart may be normal in size or moderately enlarged. The apical cardiac impulse is localized and forceful indicating left ventricular hypertrophy.

The striking sign is a loud harsh murmur continuous throughout systole and diastole (Gibson, 1900) It waxes towards the end of systole and wanes in mid-diastole. It has been described as a *'machinery' or 'train-in-the-tunnel' murmur.* Gibson's murmur is best heard in the left infraclavicular region (Fig. 39) The systolic

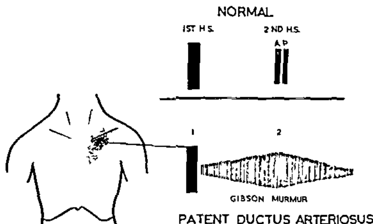



FIG 39 Auscultatory findings in patent ductus arteriosus H.S, heart sound, A, aortic element of the second heart sound, P, pulmonary element of the second heart sound, 1, first heart sound, 2, second heart sound

element of the murmur may be audible over the entire precordium A *thrill* accompanies the murmur in most cases.

The development of the continuous murmur depends on the relative pressures in the aorta and pulmonary artery. In infants and young children, where the systemic (aortic) blood pressure is relatively low, the continuous murmur may be absent and a systolic murmur only is heard at the pulmonary area. Similarly, in cases of pulmonary hypertension with reversed shunt a continuous murmur does not occur (*vide infra*). Any manoeuvre that increases the blood flow, e g. exercise, may accentuate or produce a continuous murmur.

A mid-diastolic murmur is sometimes heard in the mitral area. This is attributed to rapid filling of an enlarged left ventricle.

The second heart sound is accentuated but may be masked by the intensity of the murmur.

### Electrocardiography

In mild cases the electrocardiogram is normal. When the shunt is large there is left ventricular hypertrophy, viz. tall R waves in left ventricular leads (usually V5 and V6) and deep S waves in right ventricular leads (usually V1 and V2) In addition, left ventricular strain may occur as shown by inversion of T waves and depression of S–T segments in left ventricular leads (cf. Fig. 32).



Fig 40 X-ray of chest in patent ductus arteriosus showing cardiomegaly, increased vascularity of the lung fields, prominence of the pulmonary conus and dilatation of the pulmonary arteries

### Radiology

The characteristic radiological findings are·

1. Enlargement of the left ventricle (Fig. 40)
2. Increased vascularity of the lung fields.
3. Prominence of the pulmonary conus and dilatation of the pulmonary artery and its branches.
4. Conspicuous pulsation of the pulmonary arteries (hilar dance) on fluoroscopy. The pulsations, however, are not as marked as in atrial septal defect.

## Cardiac Catheterization

The following information may be obtained

1. *Visualization of the catheter.* The catheter is often seen to pass (Figs. 41, 42)
2. from the pulmonary r percentage oxygen



Fig 41 Postero-anterior X-ray of chest showing catheter in the axillary vein, right atrium, right ventricle, pulmonary artery and passing through a patent ductus arteriosus into the descending aorta

saturation than samples from the right ventricle, right atrium and superior vena cava (Figs. 43, 44).

3. *Pressure studies.* The pressure in the pulmonary artery is normal in mild cases and moderately raised when the shunt is large.
4. *Angiocardiography.* This is occasionally performed when the diagnosis is in doubt. The best method for demonstrating the ductus is retrograde aortography. A catheter is passed through a peripheral artery into the arch of the aorta and dye is injected.

## Clinical Presentation

The patient may present in the following ways:

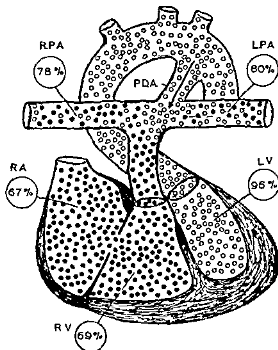
1. There are no symptoms and the diagnosis is made on routine examination.
2. The patient complains of breathlessness on exertion, palpitations and fatigue. These symptoms are more marked when there is a large shunt.



Fig 42 Lateral X-ray of chest showing catheter in the axillary vein, right atrium, right ventricle, pulmonary artery and passing through a patent ductus arteriosus into the descending aorta

3. *There may be stunting of growth and poor physical development, particularly with large shunts.*
4. *The patient may have symptoms caused by* COMPLICATIONS:
   (a) *Subacute bacterial endocarditis.* The clinical features include fatigue, pyrexia, anaemia and embolic phenomena. Vegetations tend to occur most commonly at the pulmonary end of

the ductus arteriosus, or on the wall of the left pulmonary artery opposite the opening of the ductus arteriosus. These vegetations are ideally situated for embolization to the lungs. Thus, attacks of 'pneumonia' due to pulmonary embolization may be an early manifestation of subacute bacterial endo-




percentage oxygen saturation in the pulmonary arteries, particularly the left P D A, patent ductus arteriosus, R P A, right pulmonary artery, L P A, left pulmonary artery, R A, right atrium, R V, right ventricle, L V, left ventricle

carditis, whereas petechiae, splenic infarcts and other evidence of systemic embolization tend to occur late.

(*b*) *Congestive cardiac failure.*

(*c*) *Bronchitis and bronchopneumonia.* Recurrent attacks of pulmonary infection are common. This has been attributed to

the tendency of hyperaemic lungs to react excessively to minor respiratory infections (Wood, 1956).

(*d*) *Pulmonary hypertension.*

| | S V C | RIGHT ATRIUM | RIGHT VENTRICLE | PULMONARY ARTERY |
|---|---|---|---|---|
| PATENT DUCTUS ARTERIOSUS | – | – | – | X |
| VENTRICULAR SEPTAL DEFECT | – | – | X | X |
| ATRIAL SEPTAL DEFECT | – | X | X | X |

X = Increased oxygen saturation

FIG. 44 Showing sites of increased oxygen saturation in patent ductus arteriosus, ventricular septal defect and atrial septal defect S V C, superior vena cava

PATENT DUCTUS ARTERIOSUS WITH PULMONARY HYPERTENSION

A *marked* increase in pulmonary artery pressure is sometimes found and may be due to two factors:

1. Persistence of the foetal pattern of pulmonary arterioles (see Chapter 9).
2. Reflex vasoconstriction and the development of degenerative changes in the pulmonary arterioles secondary to the increased pulmonary blood flow.

A rise in pulmonary artery pressure may have the following effects:

(*a*) The direction of flow through the ductus arteriosus is reversed and blood is shunted from the pulmonary artery to the aorta (Fig 45). Thus, unsaturated blood passes from the pulmonary artery to the aorta. As the ductus arteriosus usually joins the

aorta below the origin of the left subclavian artery, unsaturated blood enters the aorta distal to the left subclavian artery and passes to the lower limbs (Fig. 45). The head and upper limbs are supplied by branches of the aorta proximal to the entrance of the ductus arteriosus and are therefore unaffected by the shunt. Consequently, cyanosis occurs only in the lower limbs and this unequal distribution is known as *differential cyanosis*. Sometimes

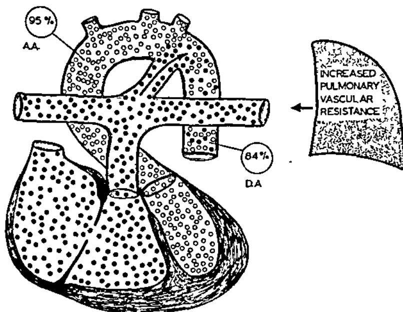



Fig. 45. Showing direction of blood flow in patent ductus arteriosus associated with pulmonary hypertension. Note: Admixture of unsaturated blood (black circles) and saturated blood (white circles) in the descending aorta. A.A., ascending aorta, D.A., descending aorta. The figures in large circles denote percentage oxygen saturation.

the ductus arteriosus enters the aorta opposite, and not distal to, the left subclavian artery. In these circumstances, unsaturated blood may pass into the left subclavian artery and the left upper limb becomes cyanosed *in addition to the lower limbs*.

(*b*) The continuous murmur disappears and a systolic murmur only is heard in the pulmonary area.

(*c*) The development of signs of pulmonary hypertension viz.:

(i) A diffuse left parasternal impulse due to right ventricular hypertrophy.
(ii) An accentuated pulmonary element of the second heart sound
(iii) Electrocardiographic evidence of right ventricular hypertrophy and strain (cf. Fig. 60, page 69).
(iv) Enlargement of the right ventricle on radiological examination.

## Treatment

Ligation or excision of the ductus arteriosus should be recommended in most cases The contraindications to operation are the presence of severe pulmonary hypertension or when the ductus arteriosus acts as a compensatory channel, e.g. when associated with Fallot's tetralogy

When subacute bacterial endocarditis complicates patent ductus arteriosus the operation must not be unduly delayed but should be preceded by an intensive course of antibiotic therapy.

Conservative treatment consists of prophylaxis of bacterial endocarditis (antibiotic cover during infections and surgical procedures) and the treatment of cardiac failure.

## General Comment

The Gibson murmur of patent ductus arteriosus may be simulated by continuous murmurs occurring in other conditions, e.g jugular venous hum (best heard in the supraclavicular fossae and varying with position of the head), arteriovenous aneurysms of the lung and rupture of an aortic sinus into the right ventricle or pulmonary artery.

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CHAPTER 6

# ATRIAL SEPTAL DEFECT

## Anatomy and Embryology

The primitive atrium is divided by a membrane—the *septum primum* —which grows from the dorsal wall of the atrium towards the endocardial bar. A temporary aperture exists between the growing edge of the septum primum and the endocardial bar This opening is called the ostium primum (Fig. 46)

Arrest in development at this stage results in a *persistent ostium primum* type of atrial septal defect (Fig. 46) A persistent ostium

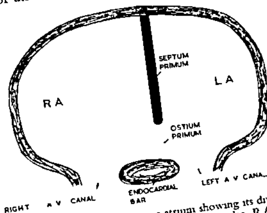



Fig 46 Transverse section of the primitive atrium showing its division into right and left atria by the septum primum A-V, atrio-ventricular, R A, right atrium, L A, left atrium

primum is often associated with lesions arising from arrested development of the endocardial bar (mitral and tricuspid valve deformities) and the interventricular septum (ventricular septal defect). The combination of persistent ostium primum with these

other malformations is known as *persistent atrioventricularis communis* or *persistent common atrioventricular canal.*

With normal development the septum primum fuses with the endocardial bar. The upper dorsal part of the septum primum

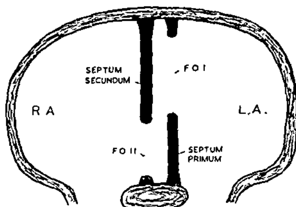



Fig. 47 Transverse section of the primitive atrium showing the formation of the septum secundum and foramina ovale R A., right atrium, L A, left atrium, F O I, foramen ovale I, F O II, foramen ovale II

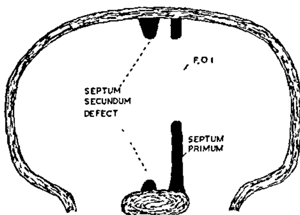



Fig 48 Diagrammatic representation showing septum secundum type of atrial septal defect F O. 1, foramen ovale 1

becomes thinned and perforate resulting in an opening—the *ostium secundum* or *foramen ovale I* (Fig. 47). Another membrane—the *septum secundum*—develops on the right side of the septum primum. It is incomplete and has an opening known as the *foramen ovale II*

(Fig. 47).* The septum secundum overlaps the foramen ovale I of the septum primum. Shortly after birth the two septa fuse to form the atrial septum proper (see also Chapter 2).

Arrested growth of the septum secundum and its failure to cover the foramen ovale I results in the *foramen secundum* type of atrial *septal defect* (Fig. 48).

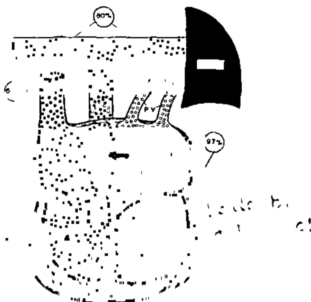



Fig. 49 Diagrammatic representation of atrial septal defect showing (*a*) flow of blood from left atrium to right atrium through the atrial septal defect, (*b*) admixture of unsaturated blood (black circles) and saturated blood (white circles) in the right atrium, right ventricle and pulmonary arteries, (*c*) average percentage oxygen saturation (figures in large circles) in the different chambers of the heart and vessels Note Increased percentage oxygen saturation in the right atrium, right ventricle and pulmonary arteries R A, right atrium, S V C, superior vena cava, R V, right ventricle, P A, pulmonary artery, A, aorta, P V, pulmonary vein

## Associated Anomalies

Cardiac malformations most commonly associated with atrial septal defect are *anomalous pulmonary venous drainage* and *pulmonary stenosis.* The combination of mitral stenosis (congenital or acquired)

* The foramina ovale I and II are sometimes collectively referred to as the foramen ovale

and atrial septal defect is known as Lutembacher's syndrome. This condition is rare.

Skeletal deformities such as arachnodactyly are often found with atrial septal defect (see Chapter 1).

## Dynamics of the Circulation

Blood is shunted from the left atrium to the right atrium through the atrial septal defect (Fig. 49). The right atrium and right ventricle thus receive an additional flow of blood Enlargement of the right atrium and right ventricle ensues. The increased right ventricular output results in a greatly increased blood flow to the lungs and

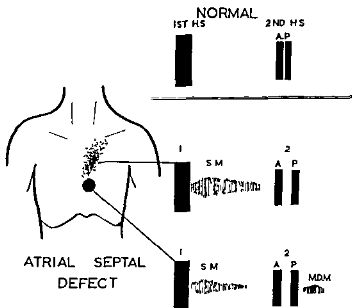



Fig 50 Auscultatory findings in atrial septal defect H S, heart sound, A, aortic element of the second heart sound, P, pulmonary element of the second heart sound, S M, systolic murmur, M D M, mid-diastolic murmur, 1, first heart sound, 2, second heart sound

dilatation of the pulmonary arteries. The amount of blood passing from the left atrium to the left ventricle is diminished and consequently the left ventricle and aorta are small The left ventricular output is less than normal.

Occasionally, the flow of blood across the defect is reversed, i e.

the flow occurs from right atrium to left atrium. This may occur with complications causing an increased pressure in the right atrium, e.g. congestive cardiac failure, pulmonary hypertension, associated pulmonary stenosis.

### Age Incidence

Most cases are found in children and young adults. The condition is, however, compatible with old age.

### Clinical Features

Patients with atrial septal defect often show poor physical development and appear thin and frail (gracile habitus). This is due to the diminished left ventricular output. Skeletal deformities, especially arachnodactyly and high-arched palate, are frequently found.

The peripheral pulse is small The blood pressure is low and the pulse pressure narrow. There is no clubbing or cyanosis.

*The heart is enlarged. There is a diffuse left parasternal impulse* due to *right ventricular hypertrophy.* The left ventricle is not enlarged. A *systolic murmur* is often heard over the second and third left intercostal spaces (Fig. 50). It may be accompanied by a *thrill.* The murmur is due to the increased flow of blood across the pulmonary valve and not to the shunt through the septal defect. A *mid-diastolic murmur* is heard at the lower left sternal border in a third of cases and is thought to be due to an increased flow through the tricuspid valve. *The second heart sound is widely split* (Fig. 50) both during expiration and inspiration.

### Electrocardiography

In most cases there is *partial or complete right bundle branch block,* i.e. the QRS complexes of right ventricular leads (usually V1 and V2) show $rSR^1$ patterns; the QRS complexes of left ventricular leads (usually V5 and V6) show deep, wide S waves (Fig. 51). In complete right bundle branch block the QRS complex is also widened to 0·12 sec. or longer.

### Radiology

The characteristic radiological findings are:

1. Gross dilatation of the pulmonary artery and its branches (Fig. 52). *On fluoroscopy there is conspicuous pulsation of the pulmonary arteries—hilar dance.*

2. Increased vascularity of the lung fields.
3. Enlargement of the right atrium and right ventricle.
4. A hypoplastic aorta and left ventricle.

### Cardiac Catheterization

The following information may be obtained:

1. *Visualization of the catheter.* The catheter may be passed through the septal defect and visualized in the left atrium.

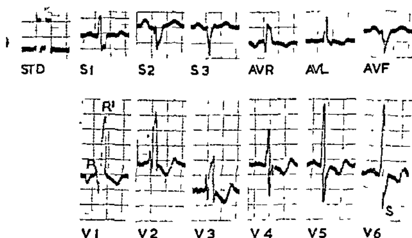



Fig 51 Electrocardiogram in atrial septal defect showing right bundle branch block (QRS complexes of leads V1, V2 and V3 show rSR[1] patterns, QRS complexes of leads V5 and V6 show deep, wide S waves)

2. *Oxygen studies.* Samples of blood from the right atrium, right ventricle and pulmonary artery show a higher percentage oxygen saturation than samples from the vena cavae (Figs. 44 and 49).
3. *Pressure studies.* The pressure in the right atrium, right ventricle and pulmonary artery is normal or slightly increased.

### Clinical Presentation

The patient may present in the following ways·

1. There are no symptoms and the diagnosis is made on routine examination. Attention to the defect may be drawn by the

presence of skeletal deformity, e.g. arachnodactyly, Marfan's syndrome

2. The patient complains of fatigue and increasing breathlessness on exertion.



FIG 52 X-ray of chest in atrial septal defect showing cardiomegaly, gross dilatation of the pulmonary arteries, prominence of the pulmonary conus and increased vascularity of the lung fields

3. The patient may have symptoms caused by COMPLICATIONS:
   (a) *Congestive cardiac failure.*
   (b) *Recurrent chest infection.*
   (c) Subacute bacterial endocarditis is very rare with atrial septal defect and if present suggests the possibility of associated congenital heart anomalies.
   (d) *Pulmonary hypertension.*

### *Atrial Septal Defect with Pulmonary Hypertension*

A marked increase in pulmonary arterial pressure is sometimes found (see Chapter 9). This, in turn, causes a rise in right ventricular and right atrial pressures. As a result, there is a reversal in the direction of flow through the septal defect and blood is now shunted from right atrium to left atrium. Unsaturated blood enters the left atrium and passes to the left ventricle, aorta and systemic circulation. Central cyanosis appears and leads to clubbing of the fingers and polycythaemia.

With the development of pulmonary hypertension the systolic murmur may disappear and the degree of right ventricular hypertrophy is increased. The interval between the aortic and pulmonary components of the widely split second heart sound is diminished and the second heart sound becomes closely split. The pulmonary element is accentuated.

*The clinical presentation is that of cyanosis and pulmonary* hypertension, a combination which may occur in other conditions, [illegible] of atrial [illegible] there-

### Treatment

Conservative therapy consists of the management of cardiac failure and the prevention and treatment of pulmonary infection.

Surgical repair of the defect is now possible under direct vision with the aid of hypothermia or extra-corporeal circulation.

### General Comment

1. *Persistent ostium primum type of atrial septal defect.* This anomaly is often associated with mitral and tricuspid valve deformities and a ventricular septal defect. The combined malformations constitute the anomaly known as *persistent common atrioventricular canal.*

   The clinical features of a persistent ostium primum defect are those of a large atrial septal defect. The murmur and thrill are pronounced. Cardiomegaly and congestive cardiac failure occur early. *In addition, there may be signs of mitral and tricuspid* incompetence. Most patients die within the first few years of life.

2. *Patent foramen ovale.* During foetal life the foramen ovale is

patent. Functional closure occurs shortly after birth due to the increased pressure in the left atrium following aeration of the lungs. The foramen ovale remains potentially patent in about 25 per cent of adults It is, however, functionally insignificant as the relatively high pressure in the left atrium keeps the walls of the foramen ovale in apposition. In conditions causing a rise of pressure in the right atrium, e.g pulmonary stenosis and pulmonary hypertension, a potentially patent foramen ovale may become functionally patent resulting in a shunt of blood from right atrium to left atrium. The shunt, however, is small and seldom causes obvious cyanosis.

3. *Paradoxical embolism.* An embolus from a peripheral vein may pass through a septal defect or foramen ovale into the systemic circulation. This is especially prone to occur when embolization *is associated with a rise in right atrial pressure.*
4. *Prognosis* In the ostium secundum type of atrial septal defect with a small to moderate shunt the prognosis is good. Complications are few and congestive cardiac failure tends to occur late.

## CHAPTER 7

# VENTRICULAR SEPTAL DEFECT

### Anatomy and Embryology

THE interventricular septum consists of two parts—the muscular and membranous septa The muscular interventricular septum develops as an outgrowth from the floor of the primitive ventricle and grows towards the endocardial bar (Fig 13). Growth of the septum ceases when it reaches the endocardial bar leaving an aperture—the *interventricular foramen*—between the muscular inter-

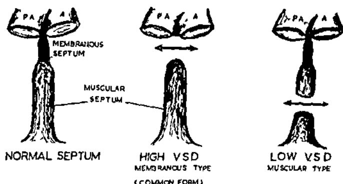



FIG 53 Diagrammatic representation showing anatomy of the interventricular septum and the membranous and muscular septal defects P A , pulmonary artery, A , aorta, V S D , ventricular septal defect

ventricular septum and the proximal part of the spiral septum The floor of this opening is formed by the endocardial bar. Closure of this opening is effected by the membranous septum which develops as an outgrowth from the endocardial bar and the endocardial ridges (see Chapter 2, page 13).

The common form of interventricular septal defect is due to non- or maldevelopment of the membranous septum. This results in a

*high or membranous ventricular septal defect.* The proximal part of this defect is formed by the roots of the pulmonary artery and aorta (Fig. 53). Less commonly, the defect is situated in the muscular septum—*low or muscular ventricular septal defect* (Fig. 53)

Ventricular septal defects vary from a few millimetres in diameter to a gap so large that there is virtually no interventricular septum (single ventricle)

## Associated Anomalies

Ventricular septal defect is often associated with other congenital cardiac anomalies, most commonly *pulmonary stenosis.*

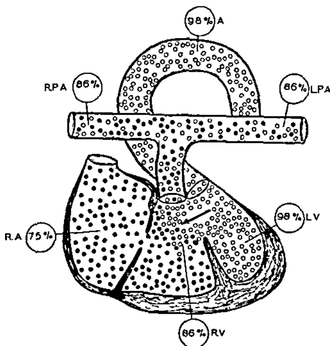



FIG 54 Diagrammatic representation of ventricular septal defect showing (a) flow [illegible] arteries and aorta Note increased percentage oxygen saturation in the right ventricle and pulmonary arteries R.A , right atrium, R.V , right ventricle, L V , left ventricle, R P.A , right pulmonary artery, L.P A., left pulmonary artery, A., aorta

## DYNAMICS OF THE CIRCULATION

In most cases the relatively high pressure in the left ventricle results in a shunt of blood through the septal defect from left ventricle to right ventricle (Fig. 54) The right ventricle thus receives an additional amount of blood The increased right ventricular output leads to a greater flow of blood to the lungs and an augmented return to the left atrium. These haemodynamics may result in left

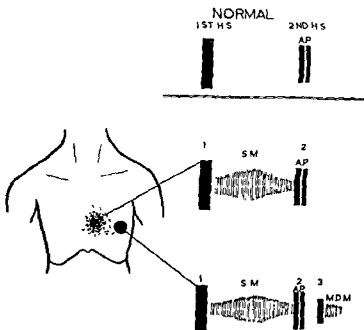



FIG 55 Auscultatory findings in ventricular septal defect H S, heart sound, A, aortic element of the second heart sound, P., pulmonary element of the second heart sound, S M, systolic murmur, M D M, mid-diastolic murmur, 1, first heart sound, 2, second heart sound, 3, third heart sound

and right ventricular hypertrophy. The left ventricle hypertrophies as a result of the strain occasioned by the shunt and the increased return of blood from the lungs. The right ventricle hypertrophies due to the extra volume of blood it receives through the septal defect.

Occasionally the flow of blood through the defect is reversed, i.e. the shunt occurs from right ventricle to left ventricle. This may

happen when the pressure in the right ventricle becomes higher than that in the left due to development of pulmonary hypertension.

### Age Incidence

Most cases are seen in the first and second decades.

### Clinical Features

The signs and symptoms vary with the size of the defect and the volume of the shunt.

The pulse and blood pressure are normal. The heart may be normal in size or moderately enlarged. A localized apical impulse is usually present indicating left ventricular hypertrophy. In addition, there may be a diffuse parasternal impulse due to right ventricular hypertrophy.

*The striking feature is a loud harsh systolic murmur* extending throughout systole The intensity of the murmur is maximal over the third and fourth interspaces at the left sternal border (Fig. 55) A *thrill* usually accompanies the murmur. A mid-diastolic murmur may be heard at the mitral area and is attributed to torrential blood flow across the mitral valve (Wood, 1950). The third heart sound is often accentuated due to rapid left ventricular filling (Fig. 55). The second heart sound may be obscured by the murmur and is normally split.

#### Electrocardiography

In mild cases the electrocardiogram is normal. With moderate or large shunts there may be evidence of both left and right ventricular hypertrophy.

Partial or complete heart block is rare.

### Radiology

In mild cases radiological examination of the chest is normal In more severe cases, however, the following features may be found:

1. Dilatation of the pulmonary artery and its branches (Fig. 56). On fluoroscopy the pulmonary arteries are seen to pulsate—hilar dance. This pulsation is not as conspicuous as in atrial septal defect.
2. Increased vascularity of the lung fields.
3. Enlargement of both ventricles, especially the left.

### Cardiac Catheterization

The following information may be obtained:

1. *Visualization of the catheter.* The catheter may be passed through the septal defect and visualized in the left ventricle.
2. *Oxygen studies.* Samples of blood from the right ventricle and pulmonary artery show a higher percentage oxygen saturation than samples from the vena cavae and right atrium (Figs. 44 and 54).
3. *Pressure studies.* The pressure in the right ventricle and pulmonary artery may be increased.



Fig. 56 X-ray of the chest in ventricular septal defect showing cardiomegaly and increased vascularity of the lung fields.

### Clinical Presentation

The patient may present in the following ways:

1. There are no symptoms and the diagnosis is made on routine examination with discovery of the systolic murmur and thrill. Roger (1879) first described this mild and asymptomatic form of ventricular septal defect and the term '*maladie de Roger*' should be confined to these cases.

2. The patient complains of palpitations and breathlessness on exertion
3. The patient may have symptoms caused by COMPLICATIONS:
   (a) *Congestive cardiac failure.*
   (b) *Recurrent chest infection.*
   (c) *Subacute bacterial endocarditis.* Vegetations form in the right ventricle, either at the site of the defect or on the wall opposite the defect where the force of the shunt is directed. [illegible]
   endocarditis.
   (d) *Pulmonary hypertension*

### Ventricular Septal Defect with Pulmonary Hypertension

In a small percentage of cases a marked increase in pulmonary arterial pressure occurs (see Chapter 9). An increase in right ventricular pressure follows This leads to reversal in the direction of flow through the defect and blood is now shunted from right ventricle to left ventricle. Unsaturated blood enters the left ventricle, aorta and systemic circulation Central cyanosis appears and leads to clubbing of the fingers and polycythaemia.

With the development of pulmonary hypertension the loud systolic murmur becomes softer or disappears, the second heart sound is accentuated and a marked degree of right ventricular hypertrophy occurs

#### Treatment

Conservative therapy includes the prophylaxis of bacterial endocarditis (antibiotic cover during infections and surgical procedures, particularly dental surgery) and the treatment of cardiac failure

Surgical repair of the defect under direct vision is now possible with the aid of extra-corporeal circulation.

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CHAPTER 8

# PULMONARY STENOSIS AND ASSOCIATED ANOMALIES

PULMONARY stenosis may occur as an isolated congenital anomaly or in association with other malformations such as ventricular septal defect, atrial septal defect and abnormalities of the aortic root * Some of the combinations are shown in Figure 57.

| PULMONARY STENOSIS<br>VALVULAR<br>INFUNDIBULAR<br>COMBINED VALVULAR AND INFUNDIBULAR<br><br>ASSOCIATED WITH | A. S. D<br><br>PATENT FORAMEN OVALE<br><br>V. S. D.<br>NORMAL AORTIC ROOT<br><br>V. S. D.<br>DEXTROPOSED AORTA<br>FALLOT'S TETRALOGY |
|---|---|

FIG. 57 Showing the common cardiac anomalies which may be associated with pulmonary stenosis A S D, atrial septal defect, V S D, ventricular septal defect

* Aortic root is the term used by Wood (1956) to describe the commencement of the aorta

## SIMPLE (ISOLATED) PULMONARY STENOSIS

### ANATOMY

There are two types of pulmonary stenosis. These may occur either singly or in combination.

1. *Valvular stenosis* The cusps of the pulmonary valve are fused to form a membranous cone or dome with a circular perforation (Fig. 58A).

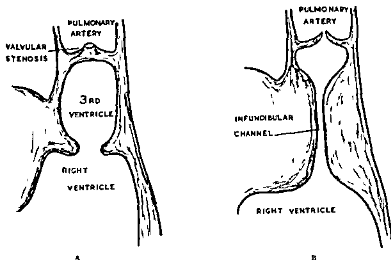



FIG 58 Diagrammatic representation showing the different types of pulmonary stenosis A Combined valvular and infundibular stenosis The infundibular stenosis in this instance is localized and demarcates the 'third ventricle' B Infundibular channel

2. *Infundibular stenosis* The outflow tract of the right ventricle may be narrowed to form a channel—the infundibular channel (Fig 58B). Alternatively, the narrowing may be localized, thus separating off a small chamber from the right ventricle This chamber is called the 'third ventricle' and is situated between the body of the right ventricle and the pulmonary valve (Fig. 58A).

### EMBRYOLOGY

Pulmonary stenosis results from abnormal development of the bulbus cordis. This structure is incorporated into the right ventricle

to form the right ventricular outflow tract and also contributes to the development of the aortic and pulmonary valves (see Chapter 2)

### Dynamics of the Circulation

The right ventricle has to pump against the obstruction caused by the pulmonary stenosis The right ventricle thus increases its force of contraction and hypertrophies. This results in a rise in right

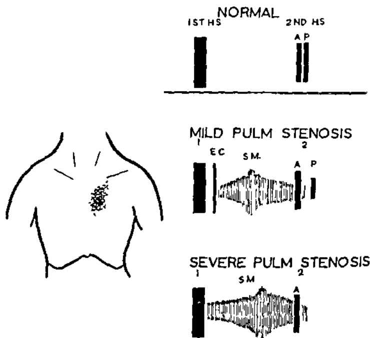



Fig 59 Auscultatory findings in pulmonary stenosis H S, heart sound, A, aortic element of the second heart sound, P, pulmonary element of the second heart sound, E C, ejection click, S M, systolic murmur, 1, first heart sound, 2, second heart sound Note Mid-systolic accentuation of the murmur in mild pulmonary stenosis and late systolic accentuation in severe stenosis.

ventricular pressure which, in turn, causes an increase in right atrial pressure and hypertrophy of the right atrium

As a result of the obstruction to the right ventricular outflow the circulation to the lungs is diminished The return of blood from the lungs is reduced and the left ventricular output is consequently low. In severe cases this may result in a fall in systemic blood pressure and compensatory peripheral vasoconstriction.

### Age Incidence

Most patients are seen during childhood and early adult life. Severe cases rarely survive the third decade.

### Clinical Features

Physical development is usually normal. There is no clubbing, polycythaemia or central cyanosis. In severe cases there may be peripheral cyanosis due to associated peripheral vasoconstriction

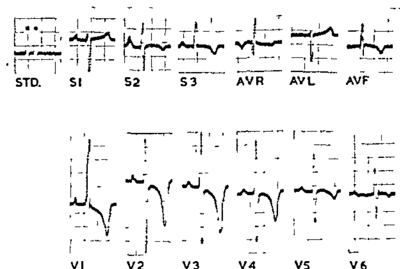



Fig. 60 Electrocardiogram in severe pulmonary stenosis showing right ventricular hypertrophy and strain (tall R waves and inverted T waves in leads V1 to V6) and right atrial hypertrophy (tall peaked P waves in leads V1 and standard 2)

The blood pressure is normal or low. An exaggerated venous pulsation in the neck—a giant *a* wave—is seen in severe cases.

There is a diffuse left parasternal impulse due to *right ventricular hypertrophy*. This may cause deformity of the chest in severe cases.

There is a *loud, harsh, protracted systolic murmur* best heard over the second left interspace (Fig. 59). A *thrill* usually accompanies the murmur. In mild cases the systolic murmur is initiated by a sharp early systolic sound—the pulmonary ejection click (Leatham and

Vogelpoel, 1954) (Fig. 59). The ejection click does not occur in severe cases. In infundibular stenosis the murmur is best heard over the third and fourth left interspaces. The pulmonary element of the second heart sound is delayed leading to wide splitting of the second sound. In severe cases the pulmonary element of the second sound occurs very late and is faint or inaudible (Fig. 59)

## ELECTROCARDIOGRAPHY

In mild cases the electrocardiogram is normal. Moderate or severe cases show the features of right ventricular hypertrophy and strain (Fig. 60)

(a) Tall R waves in right ventricular leads (usually V1 to V4)
(b) Prominent R wave in lead AVR (i.e. R wave of greater amplitude than q wave).
(c) Inverted T waves in right ventricular leads (usually leads V1 to V4)



FIG 61A X-ray of chest in valvular pulmonary stenosis showing diminished vascularity of the lung fields and post-stenotic dilatation of the pulmonary artery The angiocardiogram of this case is shown in Fig 61B

There may also be evidence of right atrial hypertrophy, viz. tall peaked P waves in right precordial leads (leads V1 and V2) and standard lead 2 (Fig. 60).

RADIOLOGY

The characteristic radiological findings are.

1 Diminished vascularity of the lung fields due to pulmonary oligaemia (Fig. 61A).



FIG 61B Angiocardiogram in a case of valvular pulmonary stenosis showing the site of the stenosis and post-stenotic dilatation of the pulmonary artery (same case as in Fig 61A) (Diagrammatic representation overleaf, Fig 61C)

2. Enlargement of the right ventricle revealed by an upturned cardiac apex in the postero-anterior view and an increased anterior bulge of the cardiac shadow in the left lateral view

3. Post-stenotic dilatation of the pulmonary artery distal to the stenosis. This dilatation appears as a prominent shadow in the

region of the pulmonary conus (Fig. 61) Post-stenotic dilatation occurs with valvular stenosis only and is due to the jet effect of the blood stream caused by the constriction.

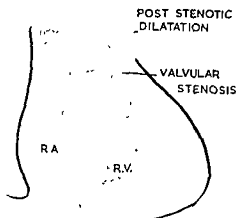



FIG 61C Diagrammatic representation of the angiocardiogram shown in Fig 61B R A, right atrium, R V, right ventricle

## CARDIAC CATHETERIZATION

The following information may be obtained·

*Pressure studies.* The pressure in the right ventricle is raised and the pulmonary arterial pressure is normal or low, i.e there is an increased pressure gradient across the pulmonary valve (Fig. 62). The more severe the pulmonary stenosis the greater is the pressure gradient

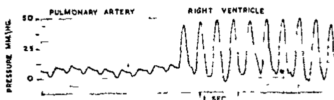



FIG. 62 Pressure tracing from a case of pulmonary stenosis showing the increased pressure gradient across the pulmonary valve as the catheter is withdrawn from the pulmonary artery into the right ventricle

*Angiocardiography*. This may be necessary to demonstrate the type of stenosis (Fig. 61B) or the presence of associated cardiac anomalies.

### Clinical Presentation

The patient may present in the following ways.

1. There are no symptoms and the diagnosis is made on routine examination with the discovery of the characteristic systolic murmur and thrill.
2. The patient complains of fatigue and breathlessness on exertion These symptoms are usually absent in mild cases
3. The patient may have symptoms caused by COMPLICATIONS
   (a) *Congestive cardiac failure.*
   (b) *Subacute bacterial endocarditis* Emboli from vegetations on the pulmonary valve travel to the lungs leading to attacks of 'pneumonia'.

### Treatment

Conservative therapy includes prophylaxis of bacterial endocarditis (antibiotic cover during infections, surgical and dental procedures) and the treatment of cardiac failure

Pulmonary valvotomy and/or infundibular resection may be performed to alleviate the obstruction. This is particularly indicated in severe cases.

## TETRALOGY OF FALLOT

### Anatomy

Fallot's tetralogy (Fallot, 1888) is the commonest form of congenital heart disease associated with cyanosis The anomaly consists of

1. PULMONARY STENOSIS.
2. HIGH MEMBRANOUS INTERVENTRICULAR SEPTAL DEFECT.
3. DEXTROPOSITION AND CONSEQUENT 'OVER-RIDING' OF THE AORTA.
4. RIGHT VENTRICULAR HYPERTROPHY.

The degree of pulmonary stenosis ranges from slight narrowing to complete atresia The stenosis is commonly infundibular in type Pure valvular stenosis or a combination of valvular and infundibular stenosis also occurs (Fig. 58).

The septal defect is situated in the membranous or sub-aortic

part of the interventricular septum (refer Chapter 7, page 60 and Fig 53).

The 'over-riding' aorta is due to an abnormality of the aortic root. Thus, the aorta is not in alignment with the left ventricular outflow tract but is displaced to the right (dextroposed) and overlies or 'over-rides' the interventricular septum from which it is separated by the high septal defect (Fig. 63).

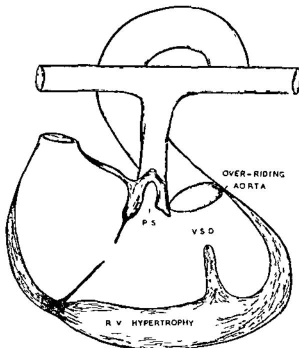



Fig 63 Diagrammatic representation of Fallot's tetralogy P S, pulmonary stenosis, V S D, ventricular septal defect, R V, right ventricular

Right ventricular hypertrophy is not a developmental abnormality but is due to the altered haemodynamics caused by the above three malformations.

## Associated Anomalies

The commonest associated congenital anomalies are *right-sided aortic arch* and *persistent left superior vena cava.*

### Embryology

Arrested development of the bulbus cordis results in malformation of the root of the pulmonary artery and its valves leading to infundibular and/or valvular stenosis.

Incomplete rotation of the spiral septum is responsible for the dextroposition and 'over-riding' of the aorta.

Failure of the aortic septum to meet the interventricular septum inevitably results in a high interventricular septal defect.

### Dynamics of the Circulation

The right ventricle pumps against the obstruction of the pulmonary stenosis Consequently, part of the right ventricular output takes the path of least resistance and is shunted into the left ventricle and aorta through the interventricular septal defect (Fig. 64). There is thus an admixture of unsaturated and saturated blood in the systemic circulation and central cyanosis occurs. Further, as a result of the shunt, part of the unsaturated blood from the right ventricle by-passes the lungs and remains unoxygenated This will aggravate the cyanosis.

Several mechanisms may compensate for this abnormal circulation

(a) *Polycythaemia.* This alleviates the anoxaemia but may aggravate the cyanosis (refer Chapter 3).

(b) *The development of an extensive collateral circulation between the branches of the aorta and the pulmonary arteries.* The principal vessels involved are the bronchial arteries. Less important collateral channels develop from mediastinal, oesophageal, internal mammary and pericardial arteries This collateral circulation enables part of the mixed saturated and unsaturated blood in the aorta to reach the lungs for oxygenation

(c) *Delayed closure of the ductus arteriosus.* In Fallot's tetralogy the ductus arteriosus forms an important channel enabling blood to be shunted from the aorta to the pulmonary artery and thence to the lungs for oxygenation This compensatory mechanism is only temporary as the ductus arteriosus eventually undergoes obliteration. Closure, however, is delayed often up to one year.

### Age Incidence

The condition is compatible with survival to middle age. Most cases, however, die before the age of 20 years.

## Clinical Features

The salient feature is *central cyanosis*. It is often present at birth but may not be obvious until early childhood This is due to relative inactivity during infancy and cyanosis may only become apparent when the child begins to walk. Cyanosis is sometimes observed for the first time during an intercurrent pulmonary infection

The intensity of the cyanosis depends on the severity of the

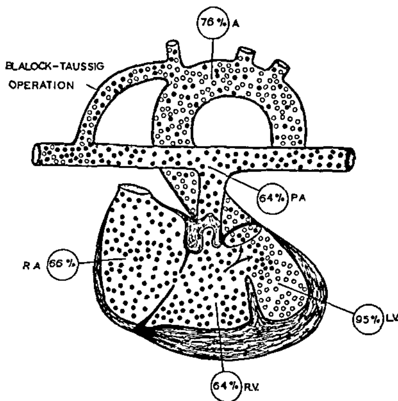



Fig 64 Diagrammatic representation of Fallot's tetralogy showing (*a*) flow of unsaturated blood (black circles) from the right ventricle to the left ventricle and aorta through a high interventricular septal defect, (*b*) admixture of unsaturated blood (black circles) and saturated blood (white circles) in the left ventricle and aorta, (*c*) average percentage oxygen saturation (figures in large circles) in the

pulmonary stenosis, the volume of the right to left shunt and the extent of the collateral circulation to the pulmonary arteries. Exercise will aggravate the cyanosis

Th

and the haematocrit reading is also high. Polycythaemia predisposes to arterial and venous thrombosis, particularly of the cerebral vessels

*Failure to thrive and stunted growth* commonly occurs in Fallot's tetralogy and all cases of congenital heart disease where cyanosis is a prominent feature.

The heart is usually normal in size. There may be a diffuse left

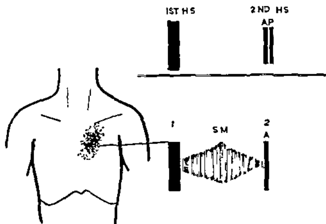



FIG 65 Auscultatory findings in Fallot's tetralogy H S, heart sound, A, aortic element of the second heart sound, P, pulmonary element of the second heart sound, S M, systolic murmur, 1, first heart sound, 2, second heart sound

parasternal impulse due to right ventricular hypertrophy. There is a *loud systolic murmur* best heard over the second and third left interspaces (Fig 65). A *systolic thrill* accompanies the murmur in most cases The thrill and murmur are due to the pulmonary stenosis and not to the shunt. The second heart sound is loud, distinct and unsplit (Fig 65) The single quality of this sound is due to the absence of any appreciable pulmonary component The clarity and accentuation of the second sound is due to the fact that the aortic root is not covered by the pulmonary artery and direct transmission of the aortic sound is thereby facilitated.

### Electrocardiography

This usually shows right ventricular hypertrophy and strain and right atrial hypertrophy (cf. Fig. 60, page 69).

### Radiology

The characteristic radiological findings are:

1. Conspicuously clear lung fields due to pulmonary oligaemia (Fig 66).
2. Hypertrophy of the right ventricle displacing the left ventricle



Fig 66 X-ray of chest in Fallot's tetralogy showing (*a*) diminished vascularity of the lung fields, (*b*) 'cocur en sabot' appearance of the cardiac silhouette, (*c*) the concavity between the aortic knuckle and ventricle.

upwards and to the left, thereby producing the appearance of a wooden shoe with an upturned toe—the 'cocur en sabot' (Fig. 66).

3. A distinct concavity between the aortic knuckle and the ventricle due to hypoplasia of the pulmonary artery (Fig. 66).

## Cardiac Catheterization

The following information may be obtained.

1. *Visualization of the catheter.* The catheter may be passed through the interventricular septal defect and visualized in the aorta or left ventricle
2. *Oxygen studies.* Samples of blood from a systemic artery, e g. femoral artery, show decreased oxygen saturation (Fig. 64) The oxygen saturation of samples from the vena cavae, right atrium, right ventricle and pulmonary artery is proportionately diminished and well below the normal venous percentage oxygen saturation range of 70 to 80 per cent (Fig. 64).
3. *Pressure studies.* The pressure in the pulmonary artery is low The pressure in the right ventricle is greatly increased and [illegible]).
4. [illegible] s simultaneous [illegible] Angiocardiography may also help determine the site and type of the pulmonary stenosis.

## Clinical Presentation

The patient may present with the following symptoms

1. *Cyanosis.* This is often present at an early age and is frequently observed by the parents.
2. *Syncope.* This is a common symptom during infancy It may be precipitated by exertion or crying and is a common cause of death in this age group.
3. *Breathlessness.* The degree of exertional dyspnoea varies with the severity of the anatomical defect. In severe cases the slightest [illegible] the arterial oxygen saturation (Lequime *et al.*, 1950)
4. Symptoms caused by COMPLICATIONS:
   (*a*) *Subacute bacterial endocarditis.*
   (*b*) *Cerebral thrombosis* is due to the polycythaemia and usually results in hemiplegia
   (*c*) *Cerebral abscess* may be caused by paradoxical embolism, i.e. infected thrombi from a peripheral vein pass through the

ventricular septal defect into the systemic and cerebral circulation. Cerebral abscess may also be due to secondary infection of a cerebral infarct following thrombosis (Campbell, 1957).

(d) Cardiac failure is rare as the septal defect acts as a safety valve to the increased pressure in the right ventricle. Further, patients usually *succumb to other complications before the* stage of cardiac failure is reached.

## Treatment

Conservative therapy includes prophylaxis of subacute bacterial endocarditis (antibiotic cover during infections, surgical and dental procedures). The attacks of dyspnoea, cyanosis and syncope, which are particularly prone to occur during infancy, are treated by the administration of oxygen and morphine, and the adoption of the knee-chest position (Taussig, 1948).

The use of surgery to correct or improve the anatomical and physiological defects in Fallot's tetralogy has resulted in considerable alleviation of symptoms and has made the prognosis more favourable. Surgery in infants carries a high mortality (30 per cent) and operation is best deferred, if possible, until the age of five or six years (Wood, 1956).

The principal surgical procedures in use are:

A. *The creation of an anastomotic channel between the aorta and* the pulmonary artery (an artificial 'ductus arteriosus'). This enables a greater volume of unsaturated blood to reach the lungs *for oxygenation. The artificial channel may be created by* anastomosing the right or left subclavian artery to the homolateral branch of the pulmonary artery. This is known as the *Blalock-Taussig operation* (Blalock and Taussig, 1945) (Fig. 64). Alternatively, the Potts-Smith operation (direct side to side anastomosis between the pulmonary artery and aorta) may be *performed* (Potts *et al.*, 1946). The Potts-Smith operation is usually carried out in infants as the technical difficulties of the Blalock-Taussig operation in this age group are considerable.

B. *Direct surgery of the pulmonary stenosis.* Infundibular resection or pulmonary valvotomy is now possible.

C. Repair of the ventricular septal defect. This is now feasible with *the aid of extracorporeal circulation.*

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## CHAPTER 9

# THE EISENMENGER SYNDROME

IN 1897 Eisenmenger described a case of ventricular septal defect associated with *central cyanosis which had been present since infancy*. It has since been shown that cases with this presentation have incomplete regression of the high intra-uterine pulmonary vascular resistance. The pulmonary arterial pressure therefore remains high and this, in turn, causes a marked rise in right ventricular pressure resulting in a right to left shunt through the ventricular septal defect (i e there is a reversal of the usual left to right shunt). This leads to central cyanosis. The combination of ventricular septal defect with cyanosis, usually dating from early childhood, is referred to as the EISENMENGER COMPLEX (Fig 67)

Other congenital anomalies which permit of communication between the venous and arterial circulations of the heart and great vessels may also be associated with persistent pulmonary hypertension and consequent reversal of the shunt across the defect. These anomalies include patent ductus arteriosus, persistent truncus arteriosus, transposition of the great vessels and atrial septal defect. These malformations, when associated with pulmonary hypertension and a reversed shunt, give rise to a clinical picture similar to the Eisenmenger complex and Wood (1952) proposed the term EISENMENGER SYNDROME to include all these conditions. The term EISENMENGER COMPLEX is reserved for those cases in which reversal of the shunt occurs through a ventricular septal defect. This condition—the Eisenmenger complex proper—is included amongst the group of defects that comprise the Eisenmenger syndrome (Fig. 67).

### ASSOCIATED ANOMALIES

A right-sided aortic arch may be associated with the Eisenmenger complex proper (Fig. 68).

## Embryology and Dynamics of the Circulation

The embryology of the congenital defects which comprise the Eisenmenger syndrome is described in the relevant chapters.

During foetal life the pulmonary vascular resistance is high. At birth, expansion of the lungs results in a diminution in pulmonary vascular resistance, and the pulmonary arterial pressure falls

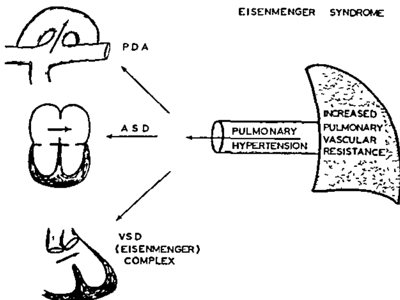



Fig. 67. Diagrammatic representation of examples of the Eisenmenger syndrome P D A, patent ductus arteriosus, A S D, atrial septal defect, V S D, ventricular septal defect

The cause of the persistent pulmonary hypertension of the Eisenmenger syndrome is still controversial. Suggested theories are.

1. Maldevelopment of the pulmonary vascular tree (Kohout *et al*, 1955)
2. Reactive pulmonary vasoconstriction. Any condition which causes a greatly increased pulmonary blood flow after birth may lead to reactive vasoconstriction of the pulmonary arterioles (Wood, 1958). Such an increase in pulmonary blood flow may occur with ventricular septal defect and patent ductus arteriosus when the *size of the defect permits of a large shunt*. Reactive vaso-

constriction prevents the transition of the pulmonary vasculature from the foetal to the extra-uterine state. This results in persistence of the high pulmonary arterial pressure and may lead to reversal of the shunt In the early stages the vasoconstriction is functional and may be relieved by vasodilators. Eventually, the pulmonary vasoconstriction is rendered permanent by the development of thrombo-obliterative changes in the pulmonary arterioles

In atrial septal defect there is little or no shunt across the defect during the first few years of life owing to the fact that both right and left ventricular pressures approximate and the resistance to diastolic filling of both ventricles is equal Hence, both atrial pressures are similar and little or no shunt occurs across the defect Thus, there is no increased pulmonary blood flow and the reactive pulmonary vasoconstriction characteristic of the Eisenmenger syndrome does not occur. The Eisenmenger syndrome is therefore less frequent with atrial septal defect than with ventricular septal defect and patent ductus arteriosus. It may, however, occur at a later stage following development of the shunt and the consequent increase in pulmonary blood flow.

## Clinical Features

Patients with the Eisenmenger syndrome have *central cyanosis* and signs of *pulmonary hypertension.*

The cyanosis is usually evenly distributed In cases of patent ductus arteriosus, however, cyanosis may be found only in the lower limbs—differential cyanosis (see also Chapter 5, page 49). Cyanosis may lead to *polycythaemia* and *clubbing of the fingers and toes*

The clinical features of pulmonary hypertension are:

(i) A diffuse left parasternal systolic impulse due to right ventricular hypertrophy.
(ii) Increased intensity of the second heart sound.
(iii) A systolic ejection click. This is a sharp sound which occurs immediately after the first heart sound and is best heard over the third left interspace. It is due to the sudden torrential flow of blood into the dilated pulmonary artery during early systole.

In the Eisenmenger syndrome the altered haemodynamics caused by the pulmonary hypertension changes the genesis of the murmurs characteristic of the various congenital anomalies. These murmurs

are therefore altered in character or absent, e g the Gibson murmur of patent ductus arteriosus may disappear and a systolic murmur only is heard. Thus, the site of the shunt in the Eisenmenger syndrome may be exceedingly difficult to diagnose clinically. Moreover, the anatomical diagnosis may be uncertain even after cardiac catheterization and angiocardiography.

### Electrocardiography

The electrocardiogram shows right ventricular hypertrophy and strain (cf. Fig. 60, page 69)



Fig 68 X-ray of the chest in a case of the Eisenmenger Complex proper showing (a) gross dilatation of the pulmonary arteries, (b) increased vascularity of the lung fields, (c) cardiomegaly Note the right-sided aortic arch which may be associated with the Eisenmenger complex proper

### Radiology

The characteristic radiological findings are

1 Gross dilatation of the pulmonary arteries (Fig. 68).

2. Increased vascularity of the lung fields.
3. Right ventricular enlargement.

### Clinical Presentation

The patient may present with the following symptoms.

1. *Cyanosis.* This may appear during early infancy or childhood in patients with ventricular septal defect and patent ductus arteriosus. Its onset in atrial septal defect is usually delayed until adulthood.
2. *Breathlessness on exertion, angina pectoris* and *syncope.*
3. Symptoms caused by COMPLICATIONS:
   (*a*) *Haemoptysis* This is commonly due to pulmonary infarction secondary to pulmonary artery thrombosis.
   (*b*) *Cardiac failure.*
   (*c*) Cerebral thrombosis and abscess (see page 79).
   (*d*) Subacute bacterial endocarditis.

### TREATMENT

Conservative therapy includes prophylaxis of subacute bacterial endocarditis, the treatment of cardiac failure and the use of anti-coagulants for the prevention and treatment of thrombotic complications

Surgical repair of the defect is strongly contraindicated in most cases of Eisenmenger syndrome and is associated with a high mortality.

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CHAPTER 10

# COMPLETE TRANSPOSITION OF THE GREAT VESSELS

## Anatomy

In complete transposition of the great vessels the aorta arises from the right ventricle and the pulmonary artery from the left ventricle This malformation is invariably associated with one or more defects

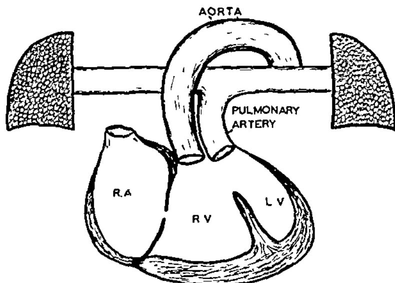



Fig 69 Diagrammatic representation of complete transposition of the great vessels associated with a ventricular septal defect R A, right atrium; R V, right ventricle, L V, left ventricle

which allow communication between the systemic and pulmonary circulations, e g. patent ductus arteriosus, atrial septal defect, patent foramen ovale and ventricular septal defect (Fig. 70).

### Embryology

The truncus arteriosus is divided into pulmonary artery and aorta by a septum—the *bulbar* or *spiral septum*. This septum spirals so that the pulmonary artery opens into the right ventricle, and the aorta into the left ventricle (see also Chapter 2, page 14). Complete transposition of the great vessels is due to failure of this septum to spiral. Consequently, the aorta opens into the right ventricle and *lies anterior to the pulmonary artery which opens into the left* ventricle.

### Dynamics of the Circulation

Blood from the right ventricle flows into the aorta and blood from the left ventricle flows into the pulmonary artery This circulatory state is obviously not compatible with life unless a communication exists between the systemic and pulmonary circulations

The course of the circulation is variable and dependent on the nature of the associated anomalies For example

Oxygenated blood returns to the left atrium from the lungs and then flows to the left ventricle and pulmonary artery. Part of this oxygenated blood escapes into the systemic circulation via

1. A patent ductus arteriosus from the pulmonary artery to aorta (Fig 70).
2. An atrial septal defect from left atrium to right atrium

Unsaturated blood returns to the right atrium and flows into the right ventricle and aorta. Some of this unsaturated blood reaches the pulmonary circulation through a ventricular septal defect (Fig 70).

*Note*: Shunts through the defects may be bidirectional, i.e the flow of blood may at times proceed in the opposite direction to that described above.

### Clinical Features

The outstanding feature is *central cyanosis*. It appears at an early age and leads to *polycythaemia* and *clubbing of the fingers and toes*.

The *heart is enlarged* and there is usually a diffuse left parasternal impulse due to right ventricular hypertrophy. The left ventricle may also be enlarged. The auscultatory findings are not diagnostic. A harsh systolic murmur may be heard at the left sternal border and the second heart sound is often accentuated

The main complaint is *breathlessness on exertion*. Physical development is usually poor. Common complications are cardiac failure, thrombotic phenomena and pulmonary infection. Most patients die at an early age; few survive beyond puberty.

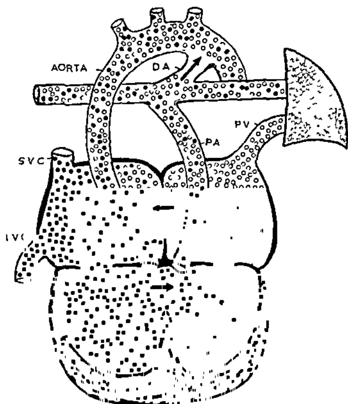



FIG 70 Example of circulatory pathway in complete transposition of the great

## ELECTROCARDIOGRAPHY

The electrocardiogram usually shows right ventricular hypertrophy and strain (cf. Fig 60, page 69).

### Radiology

The characteristic findings are:

1. Cardiomegaly involving both ventricles, principally the right.
2. Alteration in the silhouette of the great vessels. The vascular pedicle or supra-cardiac shadow is narrower than normal in the postero-anterior view (Taussig, 1938) (Fig. 71). The narrowed vascular pedicle is due to the altered anatomy of the great vessels, the pulmonary artery lying posterior to the aorta
3. Increased vascularity of the lung fields



Fig. 71 X-ray of chest in complete transposition of the great vessels showing (a) cardiomegaly, (b) increased vascularity of the lung fields, (c) narrow vascular pedicle Note: In this instance there is also a right-sided aortic arch

### Cardiac Catheterization

The significant finding on catheterization is a higher oxygen saturation of blood from the pulmonary artery compared with samples from the aorta. The catheter may be seen to pass directly from the right ventricle into the aorta.

### Treatment

Surgery of this condition is still largely in the experimental stage. The results so far are disappointing.

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CHAPTER 11

# PERSISTENT TRUNCUS ARTERIOSUS

## ANATOMY

PERSISTENT truncus arteriosus is a rare congenital anomaly in which a single large vessel replaces the normal aorta and pulmonary artery (Fig. 72). The right and left pulmonary arteries arise directly from

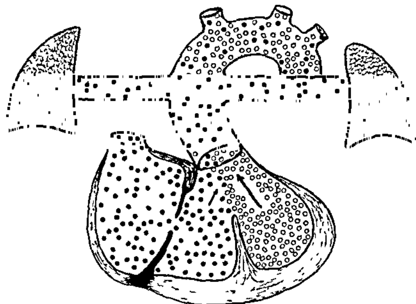


FIG 72 Diagrammatic representation showing the anatomy of persistent truncus arteriosus The pulmonary arteries arise directly from the truncus arteriosus Note the admixture of unsaturated blood (black circles) and saturated blood (white circles) in the truncus arteriosus

the truncus arteriosus. The pulmonary arteries may, however, be rudimentary or absent. A high ventricular septal defect is always found.

## Embryology

Persistent truncus arteriosus is due to failure of development of the bulbar or spiral septum which normally divides the truncus arteriosus into aorta and main pulmonary artery (see also Fig 13, page 13) The absence of the bulbar or spiral septum precludes its fusion with the interventricular septum, thereby resulting in a high ventricular septal defect.

## Dynamics of the Circulation

Blood enters the truncus arteriosus from the right and left ventricles. The truncus arteriosus therefore contains an admixture [illegible]

pulmonary arteries are rudimentary or absent the blood flow to the lungs occurs via the bronchial arteries (branches of the aorta)

## Clinical Features

The main feature is *central cyanosis.* The degree of cyanosis depends on the volume of blood reaching the lungs for oxygenation [illegible]

systolic murmur and thrill is usually present over the base of the heart The *second heart sound is loud, distinct and always single and unsplit*

Physical development is poor. The principal symptoms are breathlessness on exertion and fatigue

Complications are *cardiac failure*, pulmonary infection, thrombotic phenomena and brain abscess

## Electrocardiography

The electrocardiogram usually shows both right and left ventricular hypertrophy and strain

## Radiology

The characteristic radiological findings are

1 Gross cardiomegaly involving both ventricles.
2 Absence of the pulmonary arc This results in a concavity between the vascular shadow and the ventricle.

3 Enlargement of the supracardiac 'aortic' shadow (Fig. 73).
4. Plethoric lung fields.

The diagnosis may be confirmed by cardiac catheterization and angiocardiography

TREATMENT

In the majority of cases therapy is conservative and includes the treatment of congestive cardiac failure and pulmonary infection.



If the patient is severely cyanosed, a Blalock-Taussig operation will be of benefit and may be performed provided there is a pulmonary artery of sufficient size (as revealed by angiocardiography). This operation (see page 80 and Fig. 64) will increase the return of blood to the lungs for oxygenation.

## CHAPTER 12

# EBSTEIN'S ANOMALY

### ANATOMY

THE essential feature of this malformation is the downward displacement of part of the tricuspid valve into the right ventricle (Ebstein, 1866).

The right atrium and right ventricle are normally demarcated by

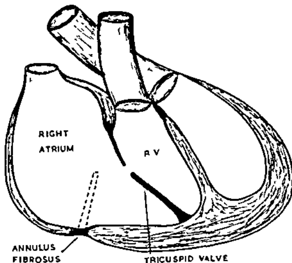



FIG 74. Diagrammatic representation of Ebstein's anomaly showing downward displacement of part of the tricuspid valve into the right ventricle The dotted line indicates the normal site of the tricuspid valve

a fibrous ring called the *annulus fibrosus* The tricuspid valve arises from this structure. In Ebstein's anomaly the anterior cusp of the tricuspid valve retains its attachment to the annulus fibrosus; the posterior and sometimes the medial cusps arise from the wall of the

right ventricle itself, resulting in their displacement downwards into the right ventricle (Fig 74). Consequently, the upper part of the right ventricle is incorporated into the right atrium. The cusps themselves may be grossly deformed and the valve may be in the form of a perforated membrane.

### Associated Anomalies

In most cases there is an associated atrial septal defect or patent foramen ovale.

### Embryology

*The cause of this malformation has not yet been established*

### Dynamics of the Circulation

The course of the circulation may be normal. When the pressure in the right atrium is increased blood may be shunted through an atrial septal defect or patent foramen ovale from the right atrium to the left atrium. An increase in right atrial pressure may result from·

(*a*) Impaired filling of the right ventricle due to the obstruction caused by the deformed valve.
(*b*) Cardiac failure or tricuspid incompetence.

### Clinical Features

The circulation is hypokinetic. The peripheral pulses are weak and the pulse pressure is small. Central cyanosis may occur but is usually only found during attacks of paroxysmal tachycardia or terminally with the development of congestive cardiac failure. Cardiomegaly may be gross The cardiac impulse is weak or impalpable. Auscultation usually reveals a triple rhythm A loud pansystolic murmur at the lower end of the sternum denotes tricuspid incompetence.

### Electrocardiography

Partial or complete right bundle branch block is frequent (cf Fig. 51, page 56). *Other disturbances of conduction may occur,* e.g. prolongation of the P-R interval. *The P waves are often tall* and pointed in standard lead 2 and leads V1 and V2, indicating right atrial hypertrophy.

## Radiology

The characteristic radiological findings are:

1. Gross cardiomegaly
2. A distinct sharp or stencilled outline of the heart The appearance is similar to that found in pericardial effusion
3. Diminished vascularity of the lung fields

## Cardiac Catheterization

This procedure is extremely hazardous in Ebstein's anomaly owing to the risk of *precipitating arrhythmias.*

## Clinical Presentation

The patient may present in the following ways

1. There may be breathlessness on exertion, palpitations and fatigue.
2. The patient may have symptoms caused by COMPLICATIONS·
   (*a*) *Paroxysmal tachycardia* Patients with Ebstein's anomaly are particularly prone to sudden attacks of paroxysmal tachycardia.
   (*b*) *Congestive cardiac failure.*

## Treatment

There is no specific treatment.

## REFERENCE

Ebstein, W (1866) 'Ueber einen sehr seltenen Fall von Insufficienz der Valvula Tricuspidalis, bedinger durch eine Angeborene hochgradige Missbildung derselben *Arch f Anat u Physiol*, 238

CHAPTER 13

# TRICUSPID ATRESIA

## Anatomy

In this condition the tricuspid valve is in the form of a small, thickened, non-perforated membrane. The right ventricle is small, rudimentary and usually filled with blood clot (Fig 75).

Tricuspid atresia is always associated with an atrial septal defect or patent foramen ovale.

## Embryology

Tricuspid atresia is due to maldevelopment of the interventricular septum and atrio-ventricular canal. The endocardial bar divides the common atrio-ventricular canal into right and left atrioventricular canals (Figs. 13, 46, 47). Normally, the interventricular septum grows towards and fuses with the endocardial bar. In tricuspid atresia the interventricular septum grows towards the right side of the endocardial bar and fuses with the margin of the right atrio-ventricular canal. As the tricuspid valve is formed from the margin of the right atrio-ventricular canal, interference with development of this canal results in tricuspid atresia.

## Dynamics of the Circulation

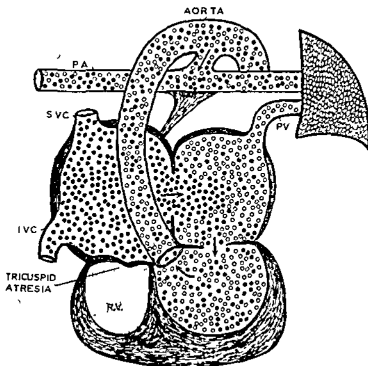
Atresia of the tricuspid valve obstructs the flow of blood from right atrium to right ventricle. Blood from the right atrium therefore flows through an atrial septal defect or patent foramen ovale into the left atrium. The left atrium thus contains an admixture of saturated and unsaturated blood which passes into the left ventricle and through the aorta into the systemic circulation (Fig 75).

Blood reaches the lungs by one or more of the following pathways:

(a) From the left ventricle to the aorta and then through a patent ductus arteriosus to the pulmonary arteries. With this type of

circulatory pathway, the main pulmonary artery is atretic (Fig. 75).

(*b*) From the left ventricle through a ventricular septal defect into




[illegible] ventricle and aorta P A, pulmonary artery, P V, pulmonary vein, S V C, superior vena cava, I V C, inferior vena cava, R V, right ventricle

the rudimentary right ventricle and thence through the main pulmonary artery to the lungs.

(*c*) From the left ventricle to the aorta and then through broncho-pulmonary anastomotic vessels to the lungs

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CHAPTER 14

# ANOMALOUS PULMONARY VENOUS DRAINAGE

In this condition one or more of the pulmonary veins drain directly or indirectly into the right atrium. When all the pulmonary veins are involved the malformation is referred to as *total anomalous pulmonary venous drainage.* More often, only part of the pulmonary venous drainage follows this abnormal course and the condition is then known as *partial anomalous venous drainage*

## TOTAL ANOMALOUS PULMONARY VENOUS DRAINAGE

### Development of the Sinus Venosus and Great Veins

The sinus venosus is the primitive chamber which receives the venous drainage of the foetus. It is situated posterior to and communicates with the primitive atrium (Fig 76). The sinus venosus consists of two lateral sacculations—the *right and left horns*—which are joined by a narrow *transverse channel* (Fig 76).

The venous return to the sinus venosus is conveyed by three paired primitive vessels

(*a*) The *vitelline veins* which drain the yolk sac.
(*b*) The *umbilical veins* which drain the placenta.
(*c*) The *cardinal veins* which drain the body of the foetus itself.

The cardinal veins comprise the *right and left anterior cardinal veins* which drain the cephalic region and the *right and left posterior cardinal veins* which drain the trunk and caudal region. These veins unite to form the *common cardinal veins* (ducts of Cuvier) which empty into the right and left horns of the sinus venosus (Fig. 76).

#### THE FATE OF THE CARDINAL VEINS AND SINUS VENOSUS

The right posterior cardinal vein eventually forms the azygos

vein which drains into the superior vena cava (Figs. 77 and 78). A transverse anastomosis develops between the two anterior cardinal veins and becomes the left innominate vein (Fig. 77). The greater part of the left anterior cardinal vein caudal to the left innominate vein normally disappears. Its remnant develops into the first left superior intercostal vein (Fig. 77). The left anterior cardinal vein may, however, fail to undergo obliteration It then

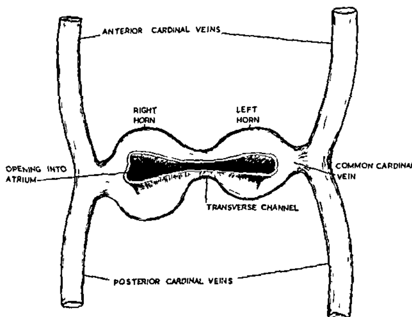



FIG 76 Diagrammatic representation showing the anatomy of the sinus venosus and cardinal veins Note The vitelline and umbilical veins which also drain into the sinus venosus have been omitted for clarity of presentation

develops into a *persistent left superior vena cava* which drains either into the left innominate vein or into the coronary sinus (Fig. 78).

The right anterior cardinal vein cephalad to the left innominate vein persists as the right innominate vein. The right anterior cardinal vein caudad to the left innominate vein becomes the superior vena cava proper which drains into the right horn of the sinus venosus (Fig. 77).

The right horn of the sinus venosus is eventually incorporated into the right atrium. The superior vena cava then drains directly

into the right atrium. The transverse channel of the sinus venosus persists as the c

the heart itself.

disappears; its

(Fig. 77).

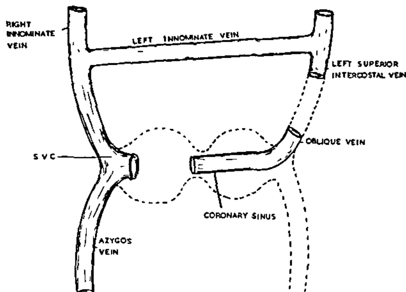



FIG 77 Diagrammatic representation showing development of the veins S V C, superior vena cava

*DEVELOPMENT OF THE PULMONARY VEINS*

The primitive lungs are drained by a surrounding venous plexus Several channels from this plexus empty into the cardinal veins These venous channels atrophy after development of the pulmonary veins proper The pulmonary vein arises as an outgrowth from that part of the primitive atrium which becomes the left atrium proper This pulmonary vein divides into left and right branches which in turn divide again forming a total of four pulmonary veins. These pulmonary veins anastomose with the pulmonary venous plexus. With growth of the atrium the pulmonary veins are drawn into the atrial wall until finally all four pulmonary veins drain directly into the left atrium.

## Embryology and Anatomy of Total Anomalous Pulmonary Venous Drainage

Complete failure of the pulmonary vein to develop results in persistence of the venous channels (anomalous pulmonary veins) which drain from the pulmonary plexus into the cardinal veins. The pulmonary venous drainage will then flow into one or more vessels which develop from these cardinal veins Thus, the anomalous

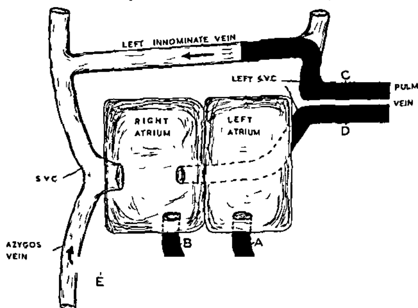


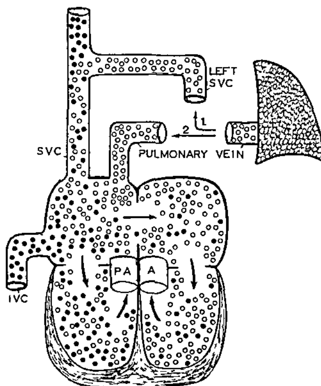

Fig 78 Diagrammatic representation showing possible pathways of total anomalous pulmonary venous drainage into the right atrium A, normal pathway, B, C, D, E, anomalous pathways (see text for explanation) S V C, superior vena cava

pulmonary veins drain into the right atrium through one or more of the following pathways:

1. Through a persistent left superior vena cava into the left innominate vein and then into the superior vena cava proper and right atrium (C in Fig. 78).
2. Through a persistent left superior vena cava into the coronary sinus and then into the right atrium (D in Fig. 78).
3. Directly into the right atrium (B in Fig. 78).
4. Through the azygos vein into the superior vena cava proper and right atrium (E in Fig. 78).

## Dynamics of the Circulation

The right atrium receives blood from both the vena cavae and the pulmonary veins (Fig. 79). An increased volume of blood thus flows into the right atrium, right ventricle and pulmonary artery. Blood can reach the left atrium only through a patent foramen ovale or an atrial septal defect. The left atrium therefore receives both saturated and unsaturated blood from the right atrium. This admixture of blood passes into the left ventricle, aorta and systemic circulation resulting in central cyanosis




A, aorta

## Clinical Features

Mild or moderate *cyanosis* is found and may be accompanied by polycythaemia and clubbing. Physical development is poor. The peripheral pulses are small in volume and the pulse pressure is narrow. There is cardiomegaly with a diffuse left parasternal



Fig 80 X-ray of chest in total anomalous pulmonary venous drainage through a persistent left superior vena cava. Note the large supracardiac vascular shadow which produces a characteristic cardiovascular silhouette resembling a 'figure-of-eight' or 'cottage-loaf'

*impulse due to right ventricular hypertrophy. Murmurs are not* characteristic.

Symptoms include breathlessness on exertion and fatigue. Recurrent respiratory infections are frequent. Cardiac failure is the commonest cause of death. Few patients survive beyond the first year of life.

### Electrocardiography

The electrocardiogram commonly shows right ventricular hypertrophy (cf. Fig. 60, page 69). There may be evidence of right atrial hypertrophy which is revealed by tall, peaked P waves in standard lead 2 and in leads V1 and V2 (cf Fig 60)

### Radiology

The characteristic findings are:

1. Enlargement of the right ventricle and right atrium
2. Increased vascularity of the lung fields and dilatation of the pulmonary arteries
3. A large ovoid supracardiac shadow caused by the dilated persistent left superior vena cava, left innominate vein and right superior vena cava. This ovoid supracardiac shadow produces a characteristic cardiovascular silhouette resembling in appearance a '*figure-of-eight*' or 'cottage-loaf (Fig 80). This sign is diagnostic of total anomalous pulmonary venous drainage *through a persistent left superior vena cava* It does not occur when the anomalous pulmonary venous drainage takes place through other channels.

### Cardiac Catheterization

The diagnosis may be confirmed by cardiac catheterization and angiocardiography.

Characteristically the percentage oxygen saturation in all four chambers of the heart is similar. The catheter may be seen to pass directly from the right atrium into an anomalous pulmonary vein

### Treatment

*Surgical treatment of total anomalous pulmonary venous drainage* is still in a development phase and is associated with a high mortality.

## PARTIAL ANOMALOUS PULMONARY VENOUS DRAINAGE

In this condition part of the pulmonary venous drainage (usually one or two pulmonary veins) drains directly into the right atrium The circulatory dynamics are similar to those of atrial septal defect which is often an associated finding Diagnosis is made by radiological examination and cardiac catheterization.

CHAPTER 15

# DEXTROCARDIA

In this condition the heart is situated predominantly in the right side of the chest with the apex pointing to the right. The position of the heart is thus a 'mirror-image' of the normal

Dextrocardia may occur as the only abnormality—*isolated dextrocardia*—or it may be associated with transposition of the abdominal viscera (*situs inversus*) Dextrocardia, particularly isolated dextrocardia, may be associated with other congenital cardiac anomalies, e.g. pulmonary stenosis, persistent truncus arteriosus and transposition of the great vessels.

Dextrocardia associated with bronchiectasis and sinusitis is known as *Kartagener's syndrome*

## Embryology

Dextrocardia is due to rotation of the primitive cardiac loop in the opposite direction to normal.

## Clinical Features

Dextrocardia does not give rise to symptoms unless there is an associated cardiac anomaly

The cardiac impulse is palpated on the right side of the chest. Likewise, the heart sounds are best heard over the right side.

## Electrocardiography

The electrocardiogram is pathognomonic (Fig. 81). The patterns are a 'mirror-image' of those found in the normal.

1. Standard lead 1 shows a deep S wave and inverted P and T waves.
2. Lead AVR shows a qR complex with an upright P wave. It is thus similar to the pattern usually found in lead AVL.
3. Lead AVL shows an rS complex and downward P and T waves

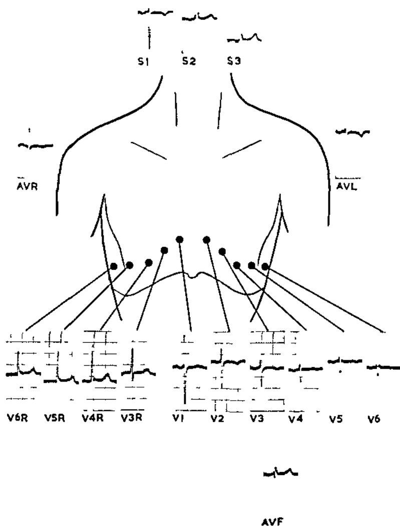



FIG 81 Electrocardiogram in dextrocardia showing 'mirror-image' patterns of the normal.

It therefore resembles the negative pattern usually found in lead AVR.

Leads taken across the left side of the chest (V1 to V6) reveal complexes with diminished amplitude of the R waves and thus resemble the normal right chest leads.

5. Leads taken across the right side of the chest (V1, V3R, V4R, V5R, V6R) show patterns which resemble those normally found in the left precordial leads.




FIG 82 X-ray of chest in dextrocardia with situs inversus Note The stomach gas shadow is situated on the right and the left dome of the diaphragm is at a higher level than the right.

## RADIOLOGY

X-ray of the chest will show the heart on the right side. With situs inversus the stomach gas shadow is also situated on the right side and the left dome of the diaphragm is at a higher level than the right (Fig. 82).

CHAPTER 16

# SOME CORRELATIVE OBSERVATIONS

## AGE INCIDENCE AND PROGNOSIS

FALLOT'S tetralogy is the commonest congenital malformation associated
in this gro
particularl
of Fallot
of cyanotic congenital heart disease die within the first decade.

## PHYSICAL DEVELOPMENT

Most patients with cyanotic congenital heart disease or large left to right shunts have poor physical development By contrast, patients with coarctation of the aorta are well developed physically.

## VENTRICULAR HYPERTROPHY

Cyanotic congenital heart disease nearly always causes right ventricular hypertrophy. The outstanding exception is tricuspid atresia which causes left ventricular hypertrophy only. Thus, the combination of cyanotic congenital heart disease with marked left ventricular hypertrophy on clinical, electrocardiographic and radiological examination, is indicative of tricuspid atresia.

In the acyanotic group, left ventricular hypertrophy is associated with coarctation of the aorta, ventricular septal defect and patent ductus arteriosus.

## THE CHARACTER OF THE SECOND HEART SOUND

A single, unsplit second heart sound is found with Fallot's tetralogy, severe pulmonary stenosis and persistent truncus arteriosus A widely split second heart sound occurs with atrial septal defect and mild pulmonary stenosis.

## Radiological Observations

Plethoric lung fields are usually found with left to right shunts, e.g. atrial septal defect, patent ductus arteriosus and ventricular septal defect. Cyanotic congenital heart anomalies associated with plethoric lung fields are transposition of the great vessels and persistent truncus arteriosus.

In pulmonary stenosis, Fallot's tetralogy and Ebstein's anomaly the lung fields are oligaemic.

Conspicuous pulsation of the pulmonary arteries—hilar dance—is a feature of *atrial septal defect* and occurs to a lesser extent in patent ductus arteriosus and ventricular septal defect.

Vigorous pulsation of the heart and aorta is found when coarctation of the aorta is associated with aortic incompetence due to bicuspid aortic valves

Diminished pulsation of the heart—a 'quiet' heart—is associated with Ebstein's anomaly.

## Complications

All cases of cyanotic congenital heart disease tend to develop polycythaemia and are therefore prone to thrombotic complications

Subacute bacterial endocarditis may complicate any congenital cardiac anomaly but is exceptionally rare with the septum secundum type of atrial septal defect.

# INDEX